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METABOLIC DISORDERS PANEL DG-5.1.0 (827 GENES)

Gene	*Twist X2 covered 10x*	*Twist X2 covered 20x*	*srWGS covered 10x*	*srWGS covered 15x*	*srWGS covered 20x*	*Associated Phenotype description and OMIM disease ID*
A4GALT	100%	100%	100%	100%	98.8%	[Blood group, P1Pk system, P(2) phenotype], 111400;NOR polyagglutination syndrome, 111400;[Blood group, P1Pk system, p phenotype], 111400
AASS	100%	100%	100%	100%	99.8%	Hyperlysinemia, 238700
ABAT	100%	100%	100%	100%	99.4%	GABA-transaminase deficiency, 613163
ABCA1	100%	100%	100%	100%	99.8%	Tangier disease, 205400;HDL deficiency, familial, 1, 604091
ABCC8	100%	100%	100%	100%	99.5%	Diabetes mellitus, permanent neonatal 3, 618857;Maturity-onset diabetes of the young, type 12, 621196;Diabetes mellitus, transient neonatal 2, 610374;Diabetes mellitus, noninsulin-dependent, 125853;Hypoglycemia of infancy, leucine-sensitive, 240800;Hyperinsulinemic hypoglycemia, familial, 1, 256450
ABCD1	89.3%	86.3%	98.7%	89.2%	70.9%	Adrenoleukodystrophy, 300100;Adrenomyeloneuropathy, adult, 300100
ABCD2	100%	100%	100%	100%	99.7%	

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ABCD3	100%	100%	100%	100%	99.5%	?Bile acid synthesis defect, congenital, 5, 616278;Oculopharyngodistal myopathy 5, 621446
ABCD4	100%	99.6%	100%	99.9%	99.3%	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ABCG5	100%	100%	100%	100%	99.5%	Sitosterolemia 2, 618666
ABCG8	100%	100%	100%	100%	99.7%	Sitosterolemia 1, 210250;{Gallbladder disease 4}, 611465
ABHD12	100%	100%	100%	100%	99.6%	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
ABHD5	100%	100%	100%	100%	99.9%	Chanarin-Dorfman syndrome, 275630
ACACA	100%	100%	100%	100%	99.7%	Acetyl-CoA carboxylase deficiency, 613933
ACAD8	100%	99.9%	100%	99.9%	99.5%	Isobutyryl-CoA dehydrogenase deficiency, 611283
ACAD9	100%	100%	100%	100%	99.7%	Mitochondrial complex I deficiency, nuclear type 20, 611126
ACADM	93.1%	91.3%	100%	100%	99.7%	Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450
ACADS	100%	100%	100%	100%	99.5%	Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470
ACADSB	100%	100%	100%	99.9%	99.7%	2-methylbutyrylglycinuria, 610006
ACADVL	100%	100%	100%	100%	99.3%	VLCAD deficiency, 201475
ACAT1	100%	100%	100%	100%	99.6%	Alpha-methylacetoacetic aciduria, 203750

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ACAT2	100%	100%	100%	100%	99.9%	?ACAT2 deficiency, 614055
ACBD5	85.6%	85.6%	100%	100%	99.5%	Retinal dystrophy with leukodystrophy, 618863
ACBD6	79.5%	79.5%	100%	99.9%	99.6%	Neurodevelopmental disorder with progressive movement abnormalities, 620785
ACO2	90.7%	86.8%	100%	100%	99.7%	Optic atrophy 9, 616289;Infantile cerebellar-retinal degeneration, 614559
ACOX1	97.9%	97.9%	100%	100%	99.7%	Mitchell syndrome, 618960;Peroxisomal acyl-CoA oxidase deficiency, 264470
ACOX2	100%	100%	100%	100%	99.7%	Bile acid synthesis defect, congenital, 6, 617308
ACSF3	94.7%	94.7%	100%	100%	99.3%	Combined malonic and methylmalonic aciduria, 614265
ACSL4	100%	100%	99.1%	91.5%	73.4%	Intellectual developmental disorder, X-linked 63, 300387
ACY1	100%	100%	100%	99.9%	99.4%	Aminoacylase 1 deficiency, 609924
ADA	85.1%	84.4%	100%	99.9%	99.5%	Adenosine deaminase deficiency, partial, 102700;Severe combined immunodeficiency due to ADA deficiency, 102700
ADCK5	93.5%	93.5%	100%	100%	99.5%	

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ADCY5	97.5%	96.9%	100%	99.9%	99.1%	Dyskinesia with orofacial involvement, autosomal dominant, 606703;Neurodevelopmental disorder with hyperkinetic movements and dyskinesia, 619651;Dyskinesia with orofacial involvement, autosomal recessive, 619647
ADK	88%	88%	100%	100%	99.8%	Hypermethioninemia due to adenosine kinase deficiency, 614300
ADSL	99.5%	97.5%	100%	100%	99.7%	Adenylosuccinase deficiency, 103050
AGA	91.6%	91.6%	100%	100%	99.5%	Aspartylglucosaminuria, 208400
AGK	91.6%	88.9%	100%	100%	100%	Cataract 38, autosomal recessive, 614691;Sengers syndrome, 212350
AGL	100%	100%	100%	100%	99.7%	Glycogen storage disease IIIa, 232400;Glycogen storage disease IIIb, 232400
AGPAT2	100%	99.9%	100%	100%	98.9%	Lipodystrophy, congenital generalized, type 1, 608594
AGPS	97.3%	97.3%	100%	100%	99.8%	Rhizomelic chondrodysplasia punctata, type 3, 600121
AGXT	97.4%	96.4%	100%	100%	99.5%	Hyperoxaluria, primary, type 1, 259900
AHCY	100%	100%	100%	99.9%	99.1%	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752

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AK1	100%	100%	100%	100%	99.8%	Anemia, congenital, nonspherocytic hemolytic, 3, adenylate kinase deficient, 612631
AK2	98.1%	93.2%	100%	100%	99.1%	Reticular dysgenesis, 267500
AKR1C1	100%	100%	100%	100%	99.5%	
AKR1D1	88.4%	88.4%	100%	100%	99.8%	Bile acid synthesis defect, congenital, 2, 235555
ALAD	100%	100%	100%	100%	99.4%	Porphyria, acute hepatic, 612740;{Lead poisoning, susceptibility to}, 612740
ALAS2	100%	100%	99%	90.9%	70.9%	Anemia, sideroblastic, 1, 300751;Protoporphyria, erythropoietic, X-linked, 300752
ALDH18A1	100%	100%	100%	100%	99.6%	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	94.4%	94.4%	100%	99.9%	99.4%	Microphthalmia, isolated 8, 615113
ALDH2	100%	99.9%	100%	100%	99.4%	{Sublingual nitroglycerin, susceptibility to poor response to}, 610251;Alcohol sensitivity, acute, 610251;{Hangover, susceptibility to}, 610251
ALDH3A2	93.5%	93.5%	100%	100%	99.7%	Sjogren-Larsson syndrome, 270200

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ALDH4A1	97.9%	96.1%	100%	100%	99.5%	Hyperprolinemia, type II, 239510
ALDH5A1	100%	100%	100%	100%	99.5%	Succinic semialdehyde dehydrogenase deficiency, 271980
ALDH6A1	95.2%	95.2%	100%	100%	99.7%	Methylmalonate semialdehyde dehydrogenase deficiency, 614105
ALDH7A1	97.2%	97.2%	100%	100%	99.8%	Epilepsy, early-onset, 4, vitamin B6-dependent, 266100
ALDOA	100%	100%	100%	100%	99.4%	Glycogen storage disease XII, 611881
ALDOB	100%	100%	100%	99.9%	99.8%	Fructose intolerance, hereditary, 229600
ALG1	100%	100%	100%	100%	99.7%	Congenital disorder of glycosylation, type Ik, 608540
ALG10	100%	100%	100%	100%	99.8%	
ALG11	91%	91%	100%	100%	99.9%	Congenital disorder of glycosylation, type Ip, 613661
ALG12	100%	100%	100%	100%	99.6%	Congenital disorder of glycosylation, type Ig, 607143
ALG13	100%	100%	99.2%	90.7%	72.3%	Developmental and epileptic encephalopathy 36, 300884
ALG14	100%	100%	100%	100%	99.8%	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

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ALG2	100%	100%	100%	100%	99.8%	Congenital disorder of glycosylation, type Ii, 607906;Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228
ALG3	100%	100%	100%	100%	99.4%	Congenital disorder of glycosylation, type Id, 601110
ALG5	91.6%	91.6%	100%	100%	99.9%	Polycystic kidney disease 7, 620056
ALG6	100%	100%	100%	100%	99.9%	Congenital disorder of glycosylation, type Ic, 603147
ALG8	78.4%	77.9%	100%	100%	99.7%	Congenital disorder of glycosylation, type Ih, 608104;Polycystic liver disease 3 with or without kidney cysts, 617874
ALG9	100%	100%	100%	99.9%	99.5%	Gillessen-Kaesbach-Nishimura syndrome, 263210;Congenital disorder of glycosylation, type Il, 608776
ALOX12B	100%	100%	100%	100%	99.5%	Ichthyosis, congenital, autosomal recessive 2, 242100
ALPL	100%	100%	100%	100%	99.3%	Odontohypophosphatasia, 146300;Hypophosphatasia, infantile, 241500;Hypophosphatasia, childhood, 241510;Hypophosphatasia, adult, 146300
AMACR	100%	100%	100%	99.8%	99.4%	Alpha-methylacyl-CoA racemase deficiency, 614307;Bile acid synthesis defect, congenital, 4, 214950
AMN	100%	100%	100%	99.9%	99.1%	Imerslund-Grasbeck syndrome 2, 618882

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AMPD1	100%	100%	100%	100%	99.7%	Myopathy due to myoadenylate deaminase deficiency, 615511
AMPD3	100%	100%	100%	100%	99.8%	[AMP deaminase deficiency, erythrocytic], 612874
AMT	100%	100%	100%	99.9%	99.3%	Glycine encephalopathy 2, 620398
AP1B1	93.7%	91.9%	100%	100%	99.6%	Keratitis-ichthyosis-deafness syndrome, autosomal recessive, 242150
AP1S1	100%	100%	100%	100%	99.2%	MEDNIK syndrome, 609313
AP3B2	100%	100%	100%	100%	99.5%	Developmental and epileptic encephalopathy 48, 617276
APOA5	100%	100%	100%	100%	99.4%	Hyperchylomicronemia, late-onset, 144650;{Hypertriglyceridemia, susceptibility to}, 145750
APOB	100%	100%	100%	100%	99.7%	Hypercholesterolemia, familial, 2, 144010;Hypobetalipoproteinemia, 615558
APOC2	100%	100%	100%	100%	98.3%	Hyperlipoproteinemia, type Ib, 207750

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APOE	93.8%	93.4%	100%	100%	99.1%	Alzheimer disease 2, 104310;Sea-blue histiocyte disease, 269600;{?Alzheimer disease, protection against, due to APOE3-Christchurch}, 607822;{Coronary artery disease, severe, susceptibility to}, 617347;Lipoprotein glomerulopathy, 611771;{?Macular degeneration, age-related}, 603075;Hyperlipoproteinemia, type III, 617347
APRT	100%	100%	100%	100%	99.1%	Adenine phosphoribosyltransferase deficiency, 614723
ARG1	93%	93%	100%	100%	99.8%	Argininemia, 207800
ARSA	100%	100%	100%	99.9%	99%	Metachromatic leukodystrophy, 250100
ARSB	100%	100%	100%	100%	99.3%	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200
ARSK	100%	100%	100%	100%	99.8%	Mucopolysaccharidosis, type X, 619698
ARV1	87.3%	86.5%	100%	100%	99.8%	Developmental and epileptic encephalopathy 38, 617020
ASAH1	100%	100%	100%	99.9%	99.3%	Spinal muscular atrophy with progressive myoclonic epilepsy, 159950;Farber lipogranulomatosis, 228000
ASL	100%	100%	100%	100%	99.1%	Argininosuccinic aciduria, 207900
ASNS	100%	100%	100%	100%	99.8%	Asparagine synthetase deficiency, 615574

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ASPA	100%	100%	100%	100%	99.8%	Canavan disease, 271900
ASS1	92.3%	90%	100%	100%	99.6%	Citrullinemia, 215700
ATIC	100%	100%	100%	100%	99.9%	AICA-ribosiduria due to ATIC deficiency, 608688
ATP1A1	97.2%	95.4%	100%	100%	99.6%	Hypomagnesemia, seizures, and impaired intellectual development 2, 618314;Charcot-Marie-Tooth disease, axonal, type 2DD, 618036
ATP6AP1	99.6%	97.5%	98.9%	89.5%	70.5%	Immunodeficiency 47, 300972
ATP6AP2	100%	100%	99.3%	91.2%	74.9%	Intellectual developmental disorder, X-linked syndromic, Hedera type, 300423;?Parkinsonism with spasticity, X-linked, 300911;Congenital disorder of glycosylation, type IIr, 301045
ATP6V0A2	100%	100%	100%	100%	99.6%	Wrinkly skin syndrome, 278250;Cutis laxa, autosomal recessive, type IIA, 219200
ATP6V1A	100%	100%	100%	100%	99.8%	Cutis laxa, autosomal recessive, type IID, 617403;Developmental and epileptic encephalopathy 93, 618012
ATP6V1E1	100%	100%	100%	100%	99.6%	Cutis laxa, autosomal recessive, type IIC, 617402

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ATP7A	95%	94.8%	99.4%	92.2%	74.7%	Occipital horn syndrome, 304150;Neuronopathy, distal hereditary motor, X-linked, 300489;Menkes disease, 309400
ATP7B	100%	100%	100%	100%	99.6%	Wilson disease, 277900
ATP8B1	100%	100%	100%	100%	99.7%	Cholestasis, progressive familial intrahepatic 1, 211600;Cholestasis, intrahepatic, of pregnancy, 1, 147480;Cholestasis, benign recurrent intrahepatic, 243300
AUH	100%	100%	100%	100%	99.7%	3-methylglutaconic aciduria, type I, 250950
B3GALNT1	100%	100%	100%	100%	100%	[Blood group, P1PK system, P(k) phenotype], 111400;[Blood group, globoside system], 615021
B3GALNT2	92.6%	92.6%	100%	100%	99.7%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 11, 615181
B3GALT6	100%	98.6%	100%	99.9%	98.6%	Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349;Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640;Al-Gazali syndrome, 609465

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B3GAT3	94.7%	93.8%	100%	100%	99.8%	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600
B3GLCT	100%	100%	100%	100%	99.9%	Peters-plus syndrome, 261540
B4GALT1	91%	88.3%	100%	100%	99.6%	Combined low LDL and fibrinogen, 620364;Congenital disorder of glycosylation, type IId, 607091
B4GALT7	86.8%	86.8%	100%	100%	99.9%	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070
B4GAT1	100%	100%	100%	100%	99.3%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287
BAAT	100%	100%	100%	100%	99.8%	Hypercholanemia, familial 3, 619232
BCAT1	97%	97%	100%	100%	99.7%	
BCAT2	100%	100%	100%	100%	99.4%	Hypervalinemia and hyperleucine-isoleucinemia, 618850
BCKDHA	86.3%	86.3%	100%	99.9%	99.3%	Maple syrup urine disease, type Ia, 248600
BCKDHB	89.9%	87.7%	100%	99.9%	99.3%	Maple syrup urine disease, type Ib, 620698
BCKDK	100%	100%	100%	100%	99.6%	Branched-chain keto acid dehydrogenase kinase deficiency, 614923
BCO1	100%	100%	100%	100%	99.6%	?Hypercarotenemia and vitamin A deficiency, autosomal dominant, 115300

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BLVRA	95.2%	95.2%	100%	100%	99.8%	Hyperbiliverdinemia, 614156
BMP2	100%	100%	100%	99.8%	97.7%	Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies 1, 617877;Brachydactyly, type A2, 112600;{HFE hemochromatosis, modifier of}, 235200
BPGM	100%	100%	100%	100%	99.9%	Erythrocytosis, familial, 8, 222800
BPNT2	94.9%	94.9%	100%	100%	99.5%	Chondrodysplasia with joint dislocations, GPAPP type, 614078
BSCL2	100%	100%	100%	100%	99.6%	Lipodystrophy, congenital generalized, type 2, 269700;Neuronopathy, distal hereditary motor, autosomal dominant 13, 619112;Silver spastic paraplegia syndrome, 270685;Encephalopathy, progressive, with or without lipodystrophy, 615924
BTD	94.2%	94.2%	100%	100%	99.9%	Biotinidase deficiency, 253260
C18orf32	100%	100%	100%	100%	99%	?Glycosylphosphatidylinositol biosynthesis defect 25, 619985
C1GALT1C1	100%	100%	99.4%	93.2%	73.1%	Hemolytic uremic syndrome, atypical, 8, with rhizomelic short stature, 301110;Tn polyagglutination syndrome, somatic, 300622
C2orf69	100%	100%	100%	100%	99.9%	Combined oxidative phosphorylation deficiency 53, 619423

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CA5A	84.7%	84.7%	100%	100%	99.6%	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751
CAD	99.7%	98.5%	100%	100%	99.4%	Developmental and epileptic encephalopathy 50, 616457
CAMLG	93.3%	93.3%	100%	100%	99.8%	?Congenital disorder of glycosylation, type IIz, 620201
CANT1	100%	100%	100%	100%	99.7%	Desbuquois dysplasia 1, 251450;Epiphyseal dysplasia, multiple, 7, 617719
CAT	98.1%	98.1%	100%	100%	99.8%	Acatalasemia, 614097
CAV1	64.8%	64.8%	100%	100%	98.8%	Lipodystrophy, congenital generalized, type 3, 612526;Pulmonary hypertension, primary, 3, 615343;Lipodystrophy, familial partial, type 7, 606721
CAVIN1	100%	100%	100%	99.6%	97.4%	Lipodystrophy, congenital generalized, type 4, 613327
CBLIF	100%	100%	100%	100%	99.8%	Intrinsic factor deficiency, 261000
CBS	87.7%	85.5%	100%	100%	99.5%	Thrombosis, hyperhomocysteinemic, 236200;Homocystinuria, B6-responsive and nonresponsive types, 236200
CD320	73.6%	73.6%	100%	100%	99.3%	Methylmalonic aciduria, transient, due to transcobalamin receptor defect, 613646
CEL	100%	100%	99.9%	99.4%	97.7%	Maturity-onset diabetes of the young, type VIII, 609812
CERKL	100%	100%	100%	100%	99.8%	Retinitis pigmentosa 26, 608380

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CERS3	100%	100%	100%	100%	99.8%	Ichthyosis, congenital, autosomal recessive 9, 615023
CETP	83.8%	83.8%	100%	100%	99.8%	[High density lipoprotein cholesterol level QTL 10], 143470;Hyperalphalipoproteinemia, 143470
CFTR	100%	100%	100%	100%	99.6%	Cystic fibrosis, 219700;Sweat chloride elevation without CF;Congenital bilateral absence of vas deferens, 277180;{Pancreatitis, hereditary}, 167800;{Bronchiectasis with or without elevated sweat chloride 1, modifier of}, 211400;{Hypertrypsinemia, neonatal}
CHIT1	98.5%	94.9%	100%	100%	99.6%	[Chitotriosidase deficiency], 614122
CHKB	100%	100%	100%	100%	99.5%	Muscular dystrophy, congenital, megaconial type, 602541
CHST11	100%	100%	100%	100%	99.7%	?Osteochondrodysplasia, brachydactyly, and overlapping malformed digits, 618167
CHST14	100%	100%	100%	99.9%	98.4%	Ehlers-Danlos syndrome, musculocontractural type 1, 601776
CHST3	100%	100%	100%	100%	98.9%	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095
CHST6	100%	100%	100%	100%	99.6%	Macular corneal dystrophy, 217800
CHSY1	100%	99.9%	100%	99.8%	99.1%	Temtamy preaxial brachydactyly syndrome, 605282

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CIAO1	100%	100%	100%	99.7%	98.8%	Multiple mitochondrial dysfunctions syndrome 10, 620960
CIDEC	100%	100%	100%	100%	99.8%	?Lipodystrophy, familial partial, type 5, 615238
CLCN7	97.5%	97.5%	100%	100%	99.7%	Hypopigmentation, organomegaly, and delayed myelination and development, 618541;Osteopetrosis, autosomal recessive 4, 611490;Osteopetrosis, autosomal dominant 2, 166600
CLN3	93.1%	93.1%	100%	100%	99.4%	Ceroid lipofuscinosis, neuronal, 3, 204200;Retinitis pigmentosa 101, 621548
CLN5	83%	83%	100%	100%	99.8%	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	95.1%	95.1%	100%	100%	99.4%	Ceroid lipofuscinosis, neuronal, 6B (Kufs type), 204300;Ceroid lipofuscinosis, neuronal, 6A, 601780
CLN8	100%	100%	100%	100%	99.5%	Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003;Ceroid lipofuscinosis, neuronal, 8, 600143
CLPB	98.2%	98.2%	100%	100%	99.7%	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%	100%	100%	100%	99.9%	
COG1	100%	100%	100%	100%	99.4%	Congenital disorder of glycosylation, type IIg, 611209

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COG2	100%	100%	100%	100%	99.4%	?Congenital disorder of glycosylation, type IIq, 617395
COG3	100%	100%	100%	100%	99.8%	Congenital disorder of glycosylation, type IIbb, 620546
COG4	97%	97%	100%	100%	99.3%	Congenital disorder of glycosylation, type IIj, 613489;Saul-Wilson syndrome, 618150
COG5	96.8%	96.8%	100%	100%	99.8%	Congenital disorder of glycosylation, type IIi, 613612
COG6	100%	100%	100%	100%	99.9%	Shaheen syndrome, 615328;Congenital disorder of glycosylation, type III, 614576
COG7	95.1%	95.1%	100%	100%	99.6%	Congenital disorder of glycosylation, type IIe, 608779
COG8	100%	100%	100%	100%	99.6%	Congenital disorder of glycosylation, type IIh, 611182
COLGALT1	96%	92.4%	100%	100%	99%	Brain small vessel disease 3, 618360
COMT	92.7%	88.6%	100%	100%	99.5%	[Catechol-O-methyltransferase activity, variation in], 621296
COQ2	96.3%	96.3%	100%	100%	99.5%	{Multiple system atrophy, susceptibility to}, 146500;Coenzyme Q10 deficiency, primary, 1, 607426
COQ4	96.3%	96.3%	100%	100%	99.6%	Coenzyme Q10 deficiency, primary, 7, 616276;Spastic ataxia 10, autosomal recessive, 620666
COQ5	94.4%	94.4%	100%	100%	99.5%	?Coenzyme Q10 deficiency, primary, 9, 619028

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COQ6	100%	100%	100%	100%	99.6%	Coenzyme Q10 deficiency, primary, 6, 614650
COQ7	100%	100%	100%	100%	99.5%	Coenzyme Q10 deficiency, primary, 8, 616733;Neuronopathy, distal hereditary motor, autosomal recessive 9, 620402
COQ8A	89.1%	87.2%	100%	99.9%	99.5%	Coenzyme Q10 deficiency, primary, 4, 612016
COQ8B	92.1%	92.1%	100%	100%	99.6%	Nephrotic syndrome, type 9, 615573
COQ9	91.9%	91.9%	100%	99.9%	99.2%	Coenzyme Q10 deficiency, primary, 5, 614654
CP	100%	100%	100%	100%	99.9%	Aceruloplasminemia, 604290
CPOX	93.4%	93.4%	100%	99.9%	99%	Coproporphyria, 121300;Harderoporphyria, 618892
CPS1	100%	100%	100%	100%	99.7%	Carbamoylphosphate synthetase I deficiency, 237300
CPT1A	100%	100%	100%	100%	99.5%	CPT deficiency, hepatic, type IA, 255120
CPT2	97.3%	96.3%	100%	100%	99.7%	{Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212;CPT II deficiency, infantile, 600649;CPT II deficiency, lethal neonatal, 608836;CPT II deficiency, myopathic, stress-induced, 255110
CRAT	99.2%	96.9%	100%	99.8%	99.1%	?Neurodegeneration with brain iron accumulation 8, 617917

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CRLS1	89.6%	84.4%	100%	99.7%	99.4%	Combined oxidative phosphorylation deficiency 57, 620167
CRPPA	100%	100%	100%	100%	99.3%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643
CSGALNACT1	100%	100%	100%	100%	99.9%	Skeletal dysplasia, mild, with joint laxity and advanced bone age, 618870
CTF1	100%	100%	100%	99.6%	96.8%	
CTH	100%	100%	100%	100%	100%	Cystathioninuria, 219500
CTNS	100%	100%	100%	100%	99.4%	Cystinosis, nephropathic, 219800;Cystinosis, ocular nonnephropathic, 219750;Cystinosis, late-onset juvenile or adolescent nephropathic, 219900;Cystinosis, atypical nephropathic, 219800
CTSA	100%	100%	100%	100%	99.5%	Galactosialidosis, 256540;Brain small vessel disease 6 with leukoencephalopathy, 621394
CTSC	94.8%	94.8%	100%	100%	99.8%	Periodontitis 1, aggressive, 170650;Haim-Munk syndrome, 245010;Papillon-Lefevre syndrome, 245000
CTSD	86%	86%	100%	100%	99.5%	Ceroid lipofuscinosis, neuronal, 10, 610127

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CTSK	100%	99.8%	100%	100%	99.7%	Pycnodysostosis, 265800
CUBN	100%	100%	100%	100%	99.8%	[Proteinuria, chronic benign], 618884;Imerslund-Grasbeck syndrome 1, 261100
CYB561	100%	100%	100%	100%	99.2%	Orthostatic hypotension 2, 618182
CYB5A	57.7%	57.7%	100%	100%	99.9%	Methemoglobinemia and ambiguous genitalia, 250790
CYB5R3	95.7%	95%	100%	100%	99.2%	Methemoglobinemia, type I, 250800;Methemoglobinemia, type II, 250800
CYP11A1	100%	98%	100%	100%	99.3%	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743
CYP11B1	100%	100%	100%	100%	99.4%	Aldosteronism, glucocorticoid-remediable, 103900;Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010
CYP11B2	100%	100%	100%	100%	99.3%	Hypoaldosteronism, congenital, due to CMO I deficiency, 203400;Aldosterone to renin ratio raised;{Low renin hypertension, susceptibility to};Hypoaldosteronism, congenital, due to CMO II deficiency, 610600
CYP17A1	100%	100%	100%	100%	99.5%	17,20-lyase deficiency, isolated, 202110;17-alpha-hydroxylase/17,20-lyase deficiency, 202110
CYP19A1	100%	100%	100%	100%	99.8%	Aromatase deficiency, 613546

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CYP1B1	100%	100%	100%	100%	99.5%	Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300;Anterior segment dysgenesis 6, multiple subtypes, 617315
CYP21A2	100%	100%	100%	100%	99.4%	Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910;Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910
CYP27A1	100%	100%	100%	100%	99.5%	Cerebrotendinous xanthomatosis, 213700
CYP27B1	100%	100%	100%	100%	99.6%	Vitamin D-dependent rickets, type I, 264700
CYP2R1	100%	100%	100%	100%	99.5%	Rickets due to defect in vitamin D 25-hydroxylation deficiency, 600081
CYP2U1	100%	100%	100%	99.9%	98.8%	Spastic paraplegia 56, autosomal recessive, 615030
CYP7B1	100%	100%	100%	100%	100%	Spastic paraplegia 5A, autosomal recessive, 270800;Bile acid synthesis defect, congenital, 3, 613812
D2HGDH	99.9%	98.4%	100%	99.9%	99.4%	D-2-hydroxyglutaric aciduria, 600721
DAO	100%	100%	100%	100%	99.5%	
DBH	100%	100%	100%	100%	99.5%	Orthostatic hypotension 1, due to DBH deficiency, 223360
DBT	100%	100%	100%	100%	99.9%	Maple syrup urine disease, type II, 620699
DCXR	100%	100%	100%	100%	99.1%	[Pentosuria], 260800

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DDC	100%	100%	100%	100%	99.7%	Aromatic L-amino acid decarboxylase deficiency, 608643
DDHD1	100%	100%	100%	100%	99.3%	Spastic paraplegia 28, autosomal recessive, 609340
DDOST	100%	100%	100%	99.9%	99.2%	Congenital disorder of glycosylation, type Ir, 614507
DEGS1	100%	100%	100%	100%	99.8%	Leukodystrophy, hypomyelinating, 18, 618404
DGAT1	100%	100%	100%	100%	99.6%	Diarrhea 7, protein-losing enteropathy type, 615863
DGKE	100%	100%	100%	100%	99.7%	{Hemolytic uremic syndrome, atypical, susceptibility to, 7}, 615008;Nephrotic syndrome, type 7, 615008
DGUOK	100%	100%	100%	100%	99.9%	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	97.8%	97.8%	100%	99.7%	98.6%	Desmosterolosis, 602398
DHCR7	96.2%	96.2%	100%	100%	99.6%	Smith-Lemli-Opitz syndrome, 270400

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DHDDS	74.4%	74.4%	100%	100%	99.8%	Developmental delay and seizures with or without movement abnormalities, 617836;?Congenital disorder of glycosylation, type 1bb, 621567;Retinitis pigmentosa 59, 613861
DHFR	100%	100%	100%	100%	99.9%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DHODH	89%	86.1%	100%	100%	99.5%	Miller syndrome, 263750
DHRSX	50%	50%	50%	50%	49.7%	Congenital disorder of glycosylation, type 1DD, 301133
DHTKD1	92.7%	92.7%	100%	100%	99.4%	?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025;Alpha-aminoadipic and alpha-ketoadipic aciduria, 204750
DLD	100%	100%	100%	100%	99.8%	Dihydrolipoamide dehydrogenase deficiency, 246900
DMGDH	98.8%	98.5%	100%	100%	99.8%	Dimethylglycine dehydrogenase deficiency, 605850
DNAJC12	100%	100%	100%	100%	99.7%	Hyperphenylalaninemia, mild, non-BH4-deficient, 617384
DNAJC19	100%	100%	100%	100%	99.9%	3-methylglutaconic aciduria, type V, 610198
DNM1L	100%	100%	100%	100%	99.9%	Optic atrophy 5, 610708;Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388

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DNM2	93.3%	91.7%	100%	99.9%	99.4%	Centronuclear myopathy 1, 160150;Charcot-Marie-Tooth disease, axonal type 2M, 606482;Charcot-Marie-Tooth disease, dominant intermediate B, 606482;Lethal congenital contracture syndrome 5, 615368
DNMT1	100%	99.3%	100%	100%	99.4%	Neuropathy, hereditary sensory, type IE, 614116;Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121
DNMT3B	100%	100%	100%	100%	99.7%	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860;Facioscapulohumeral muscular dystrophy 4, digenic, 619478
DOLK	100%	100%	100%	100%	99.6%	Congenital disorder of glycosylation, type Im, 610768
DPAGT1	100%	100%	100%	100%	99.4%	Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750;Congenital disorder of glycosylation, type Ij, 608093
DPM1	99.6%	96.9%	99.6%	98.4%	96.6%	Congenital disorder of glycosylation, type Ie, 608799
DPM2	100%	99.9%	100%	100%	99.5%	Congenital disorder of glycosylation, type Iu, 615042

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DPM3	100%	100%	100%	100%	98.8%	?Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 15, 618992;Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 15, 612937
DPYD	86.9%	86.9%	100%	100%	99.9%	Dihydropyrimidine dehydrogenase deficiency, 274270;5-fluorouracil toxicity, 274270
DPYS	100%	100%	100%	100%	99.5%	Dihydropyrimidinuria, 222748
DSE	100%	100%	100%	100%	99.7%	Ehlers-Danlos syndrome, musculocontractural type 2, 615539
DTYMK	91.4%	84.2%	100%	100%	99.1%	Neurodegeneration, childhood-onset, with progressive microcephaly, 619847
EBP	100%	100%	98.8%	88.4%	69.5%	MEND syndrome, 300960;Chondrodysplasia punctata, X-linked dominant, 302960
ECHS1	71.2%	69%	100%	100%	99.3%	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277
EDEM3	100%	100%	100%	100%	99.8%	Congenital disorder of glycosylation, type IIv, 619493
EEFSEC	100%	100%	100%	100%	98.7%	Neurodevelopmental disorder with progressive spasticity and brain abnormalities, 621102
ELOVL1	100%	100%	100%	100%	99.9%	Ichthyotic keratoderma, spasticity, hypomyelination, and dysmorphic facies, 618527

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ELOVL4	100%	100%	100%	100%	99.8%	Spinocerebellar ataxia 34, 133190;Stargardt disease 3, 600110;Ichthyosis, spastic quadriplegia, and impaired intellectual development, 614457
ENO3	100%	100%	100%	100%	98.9%	Glycogen storage disease XIII, 612932
EOGT	95.8%	92.4%	100%	100%	99.6%	Adams-Oliver syndrome 4, 615297
EPG5	100%	100%	100%	100%	99.8%	Vici syndrome, 242840;Neurodevelopmental disorder with parkinsonism or other movement abnormalities, 621506
EPHX1	91.3%	90.8%	100%	100%	99.5%	
EPHX2	100%	100%	100%	100%	99.7%	{Hypercholesterolemia, familial, due to LDLR defect, modifier of}, 143890
ETFA	82.4%	82.4%	100%	100%	99.6%	Glutaric acidemia IIA, 231680
ETFB	92.9%	92.9%	100%	100%	99.3%	Glutaric acidemia IIB, 231680
ETFDH	94.1%	92.3%	100%	100%	99.9%	Glutaric acidemia IIC, 231680
ETHE1	77.1%	77.1%	100%	100%	99.7%	Ethylmalonic encephalopathy, 602473
EXT1	100%	100%	100%	100%	99.5%	Exostoses, multiple, type 1, 133700;Chondrosarcoma, 215300
EXT2	100%	100%	100%	100%	99.6%	Seizures, scoliosis, and macrocephaly syndrome, 616682;Exostoses, multiple, type 2, 133701

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EXTL3	100%	99.9%	100%	100%	99.7%	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425
EYA1	100%	100%	100%	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	100%	100%	100%	100%	99.7%	Spastic paraplegia 35, autosomal recessive, 612319
FAH	95.2%	93.3%	100%	100%	99.5%	Tyrosinemia, type I, 276700
FAM20B	100%	100%	100%	100%	99.9%	
FAR1	91%	90.9%	100%	100%	99.9%	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154;Cataracts, spastic paraparesis, and speech delay, 619338
FBN1	100%	100%	100%	100%	99.8%	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	95.5%	94.8%	100%	100%	99.7%	Fructose-1,6-bisphosphatase deficiency, 229700

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FBP2	100%	100%	100%	100%	99.7%	?Leukodystrophy, childhood-onset, remitting, 619864
FCSK	100%	100%	100%	100%	99.6%	Congenital disorder of glycosylation with defective fucosylation 2, 618324
FDFT1	100%	98.7%	100%	100%	99.5%	Squalene synthase deficiency, 618156
FECH	100%	100%	100%	100%	99.8%	Protoporphyria, erythropoietic, 1, 177000
FH	100%	100%	100%	100%	99.8%	Leiomyomatosis and renal cell cancer, 150800;Fumarase deficiency, 606812
FKRP	100%	100%	100%	100%	99.3%	Muscular dystrophy-dystroglycanopathy (congenital with or without impaired intellectual development), 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

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FKTN	95.2%	94.9%	100%	100%	99.9%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800;Cardiomyopathy, dilated, 1X, 611615;Muscular dystrophy-dystroglycanopathy (congenital without impaired intellectual development), type B, 4, 613152
FLAD1	100%	100%	100%	100%	99.5%	Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency, 255100
FLVCR1	95%	94.6%	100%	100%	99.7%	Retinopathy-sensory neuropathy syndrome, 609033;Neurodevelopmental disorder with microcephaly, absent speech, and hypotonia, 621060
FMO3	100%	100%	100%	100%	99.9%	Trimethylaminuria, 602079
FOLR1	100%	100%	100%	100%	99.5%	Neurodegeneration due to cerebral folate transport deficiency, 613068
FTCD	100%	100%	100%	99.8%	98.7%	Glutamate formiminotransferase deficiency, 229100
FUCA1	100%	100%	100%	100%	99.5%	Fucosidosis, 230000
FUT2	100%	100%	100%	99.8%	98.4%	{Norwalk virus infection, resistance to};{Vitamin B12 plasma level QTL1}, 612542;[Bombay phenotype, digenic], 616754

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FUT6	100%	100%	100%	100%	99.5%	[Fucosyltransferase 6 deficiency], 613852
FUT8	97.2%	97.2%	100%	100%	99.8%	Congenital disorder of glycosylation with defective fucosylation 1, 618005
G6PC1	100%	100%	100%	100%	99.7%	Glycogen storage disease Ia, 232200
G6PC3	96.8%	96.8%	100%	100%	99.8%	Dursun syndrome, 612541;Neutropenia, severe congenital 4, autosomal recessive, 612541
G6PD	86.3%	86.3%	99%	89.1%	71.2%	Anemia, congenital, nonspherocytic hemolytic, 1, G6PD deficient, 300908;{Resistance to malaria due to G6PD deficiency}, 611162
GAA	100%	100%	100%	100%	99.5%	Pompe disease, late-onset, 621314;Pompe disease, infantile-onset, 232300
GAD1	100%	100%	100%	100%	99.5%	Developmental and epileptic encephalopathy 89, 619124
GALC	100%	100%	100%	100%	99.6%	Krabbe disease, 245200
GALE	100%	100%	100%	100%	99.6%	Thrombocytopenia 13, syndromic, 620776;Galactose epimerase deficiency, 230350
GALK1	92.3%	92.3%	100%	100%	99.8%	Galactokinase deficiency with cataracts, 230200
GALM	84%	84%	100%	100%	99.7%	Galactosemia IV, 618881
GALNS	100%	100%	100%	100%	99%	Mucopolysaccharidosis IVA, 253000

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GALNT14	100%	100%	100%	99.9%	99.4%	
GALNT2	100%	100%	100%	100%	99.4%	Congenital disorder of glycosylation, type IIt, 618885
GALNT3	100%	100%	100%	100%	99.8%	Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900
GALT	100%	100%	100%	100%	99.5%	Galactosemia, 230400
GAMT	97%	93.2%	100%	99.8%	98.8%	Cerebral creatine deficiency syndrome 2, 612736
GANAB	100%	100%	100%	100%	99.6%	Polycystic kidney disease 3, 600666
GATM	100%	100%	100%	100%	99.9%	Cerebral creatine deficiency syndrome 3, 612718;Fanconi renotubular syndrome 1, 134600
GBA1	100%	98.3%	100%	100%	99.5%	{Lewy body dementia, susceptibility to}, 127750;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;{Parkinson disease, late-onset, susceptibility to}, 168600
GBA2	100%	100%	100%	99.9%	99.4%	Spastic paraplegia 46, autosomal recessive, 614409
GBE1	100%	100%	100%	100%	99.7%	Glycogen storage disease IV, 232500;Polyglucosan body disease, adult form, 263570
GCDH	79.8%	79.8%	100%	99.9%	99.5%	Glutaricaciduria, type I, 231670

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GCH1	100%	100%	100%	100%	99.5%	Dystonia, DOPA-responsive, 128230;Hyperphenylalaninemia, BH4-deficient, B, 233910
GCK	100%	100%	100%	99.9%	99.2%	MODY, type II, 125851;Diabetes mellitus, permanent neonatal 1, 606176;Hyperinsulinemic hypoglycemia, familial, 3, 602485;Diabetes mellitus, noninsulin-dependent, late onset, 125853
GCLC	100%	100%	100%	100%	99.6%	{Myocardial infarction, susceptibility to}, 608446;Anemia, congenital, nonspherocytic hemolytic, 7, 230450
GCLM	93.1%	93.1%	100%	100%	100%	{Myocardial infarction, susceptibility to}, 608446
GCSH	100%	100%	100%	100%	99.9%	Multiple mitochondrial dysfunctions syndrome 7, 620423
GET3	87.9%	87.9%	100%	99.9%	98.9%	?Cardiomyopathy, dilated, 2H, 620203
GET4	100%	100%	100%	100%	99.5%	?Congenital disorder of glycosylation, type IIy, 620200
GFPT1	97.5%	97.5%	100%	100%	99.8%	Myasthenia, congenital, 12, with tubular aggregates, 610542
GFUS	100%	100%	100%	100%	99.5%	
GGPS1	100%	100%	100%	100%	99.9%	Muscular dystrophy, congenital hearing loss, and ovarian insufficiency syndrome, 619518
GK	96%	96%	99.5%	92.1%	73.6%	Glycerol kinase deficiency, 307030

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GLA	91.4%	91.4%	99.3%	91.7%	72.9%	Fabry disease, cardiac variant, 301500;Fabry disease, 301500
GLB1	100%	100%	100%	100%	99.5%	GM1-gangliosidosis, type I, 230500;GM1-gangliosidosis, type III, 230650;Mucopolysaccharidosis type IVB (Morquio), 253010;GM1-gangliosidosis, type II, 230600
GLDC	97.3%	97.3%	100%	100%	99.6%	Glycine encephalopathy1, 605899
GLRA1	100%	100%	100%	100%	99.7%	Hyperekplexia 1, 149400
GLRX5	100%	100%	100%	100%	99.7%	Anemia, sideroblastic, 3, pyridoxine-refractory, 616860;Spasticity, childhood-onset, with hyperglycinemia, 616859
GLS	95.8%	95.8%	100%	100%	99.8%	CASGID syndrome, 618339;Global developmental delay, progressive ataxia, and elevated glutamine, 618412;Developmental and epileptic encephalopathy 71, 618328
GLUD1	100%	100%	100%	100%	99.8%	Hyperinsulinism-hyperammonemia syndrome, 606762
GLUL	100%	100%	100%	99.9%	99.2%	Glutamine deficiency, congenital, 610015;Developmental and epileptic encephalopathy 116, 620806
GLYCTK	100%	100%	100%	99.9%	99.3%	D-glyceric aciduria, 220120
GM2A	100%	100%	100%	100%	99.7%	GM2-gangliosidosis, AB variant, 272750

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GMPPA	100%	100%	100%	100%	98.8%	Alacrima, achalasia, and impaired intellectual development syndrome, 615510
GMPPB	100%	100%	100%	100%	99.7%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352;Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 14, 615351;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350
GMPS	100%	100%	100%	100%	99.9%	
GNE	100%	100%	100%	100%	99.8%	Sialuria, 269921;Thrombocytopenia 12 with or without myopathy, 620757;Nonaka myopathy, 605820
GNMT	100%	100%	100%	99.9%	98.2%	Glycine N-methyltransferase deficiency, 606664
GNPAT	100%	100%	100%	100%	100%	Rhizomelic chondrodysplasia punctata, type 2, 222765
GNPNAT1	100%	100%	100%	99.9%	99.3%	?Rhizomelic dysplasia, Ain-Naz type, 619598
GNPTAB	100%	100%	100%	100%	99.8%	Mucolipidosis III alpha/beta, 252600;Mucolipidosis II alpha/beta, 252500
GNPTG	90.1%	90.1%	100%	99.8%	98.5%	Mucolipidosis III gamma, 252605
GNS	94.3%	94.3%	100%	100%	99.6%	Mucopolysaccharidosis type IIID, 252940

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GOLGA2	100%	100%	100%	99.9%	99.1%	Developmental delay with hypotonia, myopathy, and brain abnormalities, 620240
GOSR2	100%	100%	100%	100%	99.5%	Epilepsy, progressive myoclonic 6, 614018;Muscular dystrophy, congenital, with or without seizures, 620166
GOT1	100%	100%	100%	100%	100%	Aspartate aminotransferase, serum level of, QTL1, 614419
GOT2	92.8%	92.8%	100%	100%	99.5%	Developmental and epileptic encephalopathy 82, 618721
GPAA1	88.5%	88.5%	100%	100%	99.4%	Glycosylphosphatidylinositol biosynthesis defect 15, 617810
GPD1	100%	100%	100%	100%	99.7%	Hypertriglyceridemia, transient infantile, 614480
GPD1L	100%	100%	100%	100%	99.4%	Brugada syndrome 2, 611777
GPHN	95.9%	95.9%	100%	100%	99.8%	Molybdenum cofactor deficiency C, 615501
GPI	100%	100%	100%	100%	99.4%	Anemia, congenital, nonspherocytic hemolytic, 4, glucose phosphate isomerase deficient, 613470
GPIHBP1	94.6%	90.9%	100%	100%	99.9%	Hyperlipoproteinemia, type 1D, 615947
GPT2	92.9%	92.9%	100%	100%	99.7%	Neurodevelopmental disorder with microcephaly and spastic paraplegia, 616281
GPX1	100%	100%	100%	100%	98.9%	Hemolytic anemia due to glutathione peroxidase deficiency, 614164

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GRHPR	86.7%	86.6%	100%	100%	99.7%	Hyperoxaluria, primary, type II, 260000
GSS	100%	100%	100%	99.9%	99.5%	Anemia, congenital, nonspherocytic hemolytic, 6, glutatione synthetase deficient, 231900;Glutathione synthetase deficiency, 266130
GUSB	98.4%	98.4%	100%	100%	99.1%	Mucopolysaccharidosis VII, 253220
GYG1	93.7%	93.7%	100%	100%	100%	?Glycogen storage disease XV, 613507;Polyglucosan body myopathy 2, 616199
GYS1	100%	100%	100%	100%	99.5%	Glycogen storage disease 0, muscle, 611556
GYS2	100%	100%	100%	100%	99.8%	Glycogen storage disease 0, liver, 240600
H6PD	100%	100%	100%	100%	99.5%	Cortisone reductase deficiency 1, 604931
HADH	87.7%	87.7%	100%	100%	99.6%	Hyperinsulinemic hypoglycemia, familial, 4, 609975;3-hydroxyacyl-CoA dehydrogenase deficiency, 231530
HADHA	96.1%	95.3%	100%	100%	99.6%	HELLP syndrome, maternal, of pregnancy, 609016;LCHAD deficiency, 609016;Mitochondrial trifunctional protein deficiency 1, 609015;Fatty liver, acute, of pregnancy, 609016
HADHB	94.6%	94.6%	100%	100%	99.7%	Mitochondrial trifunctional protein deficiency 2, 620300
HAGH	100%	100%	100%	99.9%	99.4%	[Glyoxalase II deficiency], 614033

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HCFC1	100%	100%	98.5%	88.6%	69.8%	Methylmalonic aciduria and homocysteinemia, cblX type, 309541
HEXA	100%	100%	100%	100%	99.4%	[Hex A pseudodeficiency], 272800;GM2-gangliosidosis, several forms, 272800;Tay-Sachs disease, 272800
HEXB	100%	100%	100%	99.9%	99.5%	Sandhoff disease, infantile, juvenile, and adult forms, 268800
HFE	100%	99.7%	100%	100%	99.4%	Hemochromatosis, type 1, 235200
HGD	90.1%	90%	100%	100%	99.9%	Alkaptonuria, 203500
HGSNAT	83.5%	83.5%	100%	100%	99.8%	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930;Retinitis pigmentosa 73, 616544
HIBADH	73.7%	73.7%	100%	100%	99.8%	
HIBCH	100%	100%	100%	100%	100%	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620
HK1	98.2%	96.5%	100%	99.9%	99.5%	Anemia, congenital, nonspherocytic hemolytic, 5, hexokinase deficient, 235700;Retinitis pigmentosa 79, 617460;Neuropathy, hereditary motor and sensory, Russe type, 605285;Neurodevelopmental disorder with visual defects and brain anomalies, 618547
HLCS	100%	100%	100%	100%	99.2%	Holocarboxylase synthetase deficiency, 253270

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HMBS	100%	100%	100%	100%	99.9%	Leukoencephalopathy, porphyria-related, 620711;Encephalopathy, porphyria-related, 620704;Porphyria, acute intermittent, nonerythroid variant, 176000;Porphyria, acute intermittent, 176000
HMGCL	76.2%	76.2%	100%	100%	99.5%	HMG-CoA lyase deficiency, 246450
HMGCR	100%	100%	100%	100%	99.7%	Muscular dystrophy, limb-girdle, autosomal recessive 28, 620375;[Statins, response to], 620410;[Low density lipoprotein cholesterol level QTL 3], 620410
HMGCS2	97.5%	97.5%	100%	100%	99.2%	HMG-CoA synthase-2 deficiency, 605911
HMOX1	100%	100%	100%	100%	99.5%	Heme oxygenase-1 deficiency, 614034;{Pulmonary disease, chronic obstructive, susceptibility to}, 606963
HNF1A	100%	100%	100%	100%	99.5%	Hepatic adenoma, somatic, 142330;Diabetes mellitus, insulin-dependent, 20, 612520;{Diabetes mellitus, noninsulin-dependent, 2}, 125853;MODY, type III, 600496;{Diabetes mellitus, insulin-dependent}, 222100;Renal cell carcinoma, 144700

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HNF4A	100%	100%	100%	100%	99.3%	Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young, 616026;{Diabetes mellitus, noninsulin-dependent}, 125853;MODY, type I, 125850
HOGA1	100%	100%	100%	100%	99.5%	Hyperoxaluria, primary, type III, 613616
HPD	93.7%	90.9%	100%	99.9%	98.9%	Hawkinsinuria, 140350;Tyrosinemia, type III, 276710
HPDL	100%	100%	100%	100%	99.8%	Neurodevelopmental disorder with progressive spasticity and brain white matter abnormalities, 619026;Spastic paraplegia 83, autosomal recessive, 619027
HPRT1	87.6%	87.4%	99%	91.6%	74%	Hyperuricemia, HPRT-related, 300323;Lesch-Nyhan syndrome, 300322
HS2ST1	100%	100%	100%	100%	99.7%	Neurofacioskeletal syndrome with or without renal agenesis, 619194
HS6ST1	100%	100%	100%	100%	98.6%	{Hypogonadotropic hypogonadism 15 with or without anosmia}, 614880
HS6ST2	100%	100%	98.8%	87.6%	70%	?Paganini-Miozzo syndrome, 301025
HSD11B1	100%	100%	100%	100%	99.9%	Cortisone reductase deficiency 2, 614662
HSD11B2	100%	100%	100%	100%	99.4%	Apparent mineralocorticoid excess, 218030
HSD17B10	100%	100%	99.5%	91%	71.8%	HSD10 mitochondrial disease, 300438

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HSD17B3	100%	100%	100%	100%	99.8%	Pseudohermaphroditism, male, with gynecomastia, 264300
HSD17B4	100%	100%	100%	100%	99.9%	D-bifunctional protein deficiency, 261515;Perrault syndrome 1, 233400
HSD3B2	100%	100%	100%	100%	99.6%	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency, 201810
HSD3B7	100%	100%	100%	100%	99.5%	Bile acid synthesis defect, congenital, 1, 607765
HTRA2	100%	100%	100%	100%	99.5%	{Parkinson disease 13}, 610297;3-methylglutaconic aciduria, type VIII, 617248
HYAL1	100%	100%	100%	99.9%	98.9%	Mucopolysaccharidosis type IX, 601492
IDH2	89.7%	89.5%	100%	100%	99.4%	D-2-hydroxyglutaric aciduria 2, 613657
IDH3B	100%	100%	100%	100%	99%	Retinitis pigmentosa 46, 612572
IDI1	100%	100%	100%	100%	99.5%	
IDS	100%	100%	99.4%	91.4%	72.9%	Mucopolysaccharidosis II, 309900
IDUA	96%	96%	100%	99.8%	98.7%	Mucopolysaccharidosis Is, 607016;Mucopolysaccharidosis Ih/s, 607015;Mucopolysaccharidosis Ih, 607014
IMPDH1	100%	100%	100%	100%	99.5%	Retinitis pigmentosa 10, 180105;Leber congenital amaurosis 11, 613837

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INPP5E	100%	100%	100%	99.8%	98.1%	Impaired intellectual development, truncal obesity, retinal dystrophy, and micropenis syndrome, 610156;Joubert syndrome 1, 213300
INPPL1	100%	100%	100%	99.9%	99.4%	Opsismodysplasia, 258480
INSR	100%	100%	100%	100%	99.5%	Rabson-Mendenhall syndrome, 262190;Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549;Donohue syndrome, 246200;Hyperinsulinemic hypoglycemia, familial, 5, 609968
IREB2	98.6%	98.6%	100%	100%	99.8%	Neurodegeneration, early-onset, with choreoathetoid movements and microcytic anemia, 618451
ITCH	92.6%	92.6%	100%	100%	99.8%	Autoimmune disease, multisystem, with facial dysmorphism, 613385
ITPA	76.9%	76.9%	100%	99.9%	99.4%	[Inosine triphosphatase deficiency], 613850;Developmental and epileptic encephalopathy 35, 616647
IVD	90%	90%	100%	100%	99.6%	Isovaleric acidemia, 243500
JAGN1	100%	100%	100%	100%	99.6%	Neutropenia, severe congenital, 6, autosomal recessive, 616022
KCNA2	100%	100%	100%	100%	99.3%	Developmental and epileptic encephalopathy 32, 616366

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KCNJ11	100%	100%	100%	100%	99.5%	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
KGD4	100%	100%	100%	99.8%	99.5%	
KMT2A	99.2%	99.2%	100%	100%	99.5%	Wiedemann-Steiner syndrome, 605130
KMT2D	100%	100%	100%	100%	99.2%	Branchial arch abnormalities, choanal atresia, athelia, hearing loss, and hypothyroidism syndrome, 620186;Kabuki syndrome 1, 147920
L2HGDH	92.8%	92.8%	100%	100%	99.6%	L-2-hydroxyglutaric aciduria, 236792
LAMP2	70.7%	70.7%	98.9%	91%	71.6%	Danon disease, 300257
LARGE1	96.9%	96.9%	100%	100%	99.6%	Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 6, 608840;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154
LCAT	100%	100%	100%	99.9%	99%	Fish-eye disease, 136120;Norum disease, 245900
LCT	100%	100%	100%	100%	99.5%	Lactase deficiency, congenital, 223000
LDHA	100%	100%	100%	100%	99.6%	Glycogen storage disease XI, 612933

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LDHB	100%	100%	100%	100%	99.5%	[Lactate dehydrogenase-B deficiency], 614128
LDLR	100%	100%	100%	100%	99.1%	LDL cholesterol level QTL2, 143890;Hypercholesterolemia, familial, 1, 143890
LDLRAP1	93.2%	90.2%	100%	100%	99.2%	Hypercholesterolemia, familial, 4, 603813
LFNG	99.9%	97.1%	100%	99.9%	98.8%	Spondylocostal dysostosis 3, autosomal recessive, 609813
LIAS	100%	100%	100%	100%	99.9%	Hyperglycinemia, lactic acidosis, and seizures, 614462
LIPA	95.6%	95.2%	100%	100%	99.9%	Wolman disease, 620151;Cholesteryl ester storage disease, 278000
LIPC	100%	100%	100%	100%	99.6%	{Diabetes mellitus, noninsulin-dependent}, 125853;Hepatic lipase deficiency, 614025;[High density lipoprotein cholesterol level QTL 12], 612797
LIPE	100%	100%	100%	100%	99.4%	Lipodystrophy, familial partial, type 6, 615980
LIPT1	100%	100%	100%	100%	99.8%	Lipoyltransferase 1 deficiency, 616299
LIPT2	100%	100%	100%	100%	99%	Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities, 617668
LMBRD1	86.4%	86.4%	100%	99.9%	99.6%	Methylmalonic aciduria and homocystinuria, cblF type, 277380
LMF1	91.5%	91.5%	100%	100%	99.8%	Lipase deficiency, combined, 246650

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LMNA	100%	100%	100%	99.9%	99.3%	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	92.9%	92.9%	100%	99.8%	98.3%	Microcephaly 27, primary, autosomal dominant, 619180;?Epilepsy, progressive myoclonic, 9, 616540;{Lipodystrophy, partial, acquired, susceptibility to}, 608709
LPIN1	94%	94%	100%	100%	99.7%	Myoglobinuria, acute recurrent, autosomal recessive, 268200
LPIN2	99.3%	99.2%	100%	99.9%	99.4%	Majeed syndrome, 609628
LPL	100%	100%	100%	100%	99.6%	Lipoprotein lipase deficiency, 238600;[High density lipoprotein cholesterol level QTL 11], 238600;Combined hyperlipidemia, familial, 144250

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LRAT	100%	100%	100%	100%	99.4%	Leber congenital amaurosis 14, 613341;Retinal dystrophy, early-onset severe, 613341;Retinitis pigmentosa, juvenile, 613341
LTC4S	100%	100%	99.8%	99.2%	98.3%	Leukotriene C4 synthase deficiency, 614037
LYSET	100%	100%	100%	100%	99.7%	Dysostosis multiplex, Ain-Naz type, 619345
LYST	99.5%	99.5%	100%	100%	99.9%	Chediak-Higashi syndrome, 214500
MAGT1	93.8%	93.8%	99%	88.7%	70.8%	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853;Congenital disorder of glycosylation, type Icc, 301031
MAN1B1	99.8%	98.1%	100%	100%	99.6%	Rafiq syndrome, 614202
MAN2A2	100%	100%	100%	100%	99.4%	
MAN2B1	100%	100%	100%	100%	99.1%	Mannosidosis, alpha-, types I and II, 248500
MAN2B2	89.8%	87.3%	100%	100%	99.6%	Congenital disorder of glycosylation type 1EE with or without immunodeficiency, 621140
MAN2C1	98.4%	98.4%	100%	99.9%	99.5%	Congenital disorder of deglycosylation 2, 619775
MANBA	99.4%	97.5%	100%	100%	99.5%	Mannosidosis, beta, 248510
MAOA	100%	100%	99.6%	92.1%	73.7%	Brunner syndrome, 300615

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MAT1A	100%	99.9%	100%	99.9%	99.4%	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850;Methionine adenosyltransferase deficiency, autosomal recessive, 250850
MBOAT7	100%	100%	100%	100%	99.6%	Intellectual developmental disorder, autosomal recessive 57, 617188
MBTPS1	100%	100%	100%	100%	99.4%	Spondyloepiphyseal dysplasia, Kondo-Fu type, 618392;CAOP syndrome, 621252
MCCC1	94.5%	94.5%	100%	100%	99.8%	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200
MCCC2	93.5%	93.5%	100%	100%	99.9%	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210
MCEE	100%	100%	100%	100%	99.5%	Methylmalonyl-CoA epimerase deficiency, 251120
MCOLN1	98.3%	95.7%	100%	100%	99.7%	Lisch epithelial corneal dystrophy, 620763;Mucolipidosis IV, 252650
MDH1	88.5%	88.5%	100%	100%	99.8%	?Developmental and epileptic encephalopathy 88, 618959
ME2	94.1%	94.1%	100%	100%	99.9%	
MFSD2A	100%	100%	100%	100%	99.6%	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain abnormalities, 616486

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MFSD8	100%	100%	100%	100%	100%	Macular dystrophy with central cone involvement, 616170;Ceroid lipofuscinosis, neuronal, 7, 610951
MGAT2	100%	100%	100%	99.8%	99%	Congenital disorder of glycosylation, type IIa, 212066
MINPP1	100%	100%	100%	100%	99.9%	{Thyroid carcinoma, follicular}, 188470;Pontocerebellar hypoplasia, type 16, 619527
MLYCD	100%	100%	100%	100%	99.4%	Malonyl-CoA decarboxylase deficiency, 248360
MMAA	100%	100%	100%	100%	99.8%	Methylmalonic aciduria, vitamin B12-responsive, cblA type, 251100
MMAB	91.8%	91.8%	100%	99.9%	99.7%	Methylmalonic aciduria, vitamin B12-responsive, cblB type, 251110
MMACHC	100%	100%	100%	100%	99.5%	Methylmalonic aciduria and homocystinuria, cblC type, 277400
MMADHC	79.3%	79.3%	100%	100%	99.7%	Methylmalonic aciduria and homocystinuria, cblD type, 277410;Methylmalonic aciduria, cblD type, 620953;Homocystinuria-megaloblastic anemia, cblD type, 620952
MMS19	96.5%	96.5%	100%	100%	99.7%	
MMUT	100%	100%	100%	100%	99.7%	Methylmalonic aciduria, mut(0) type, 251000
MOCOS	97.6%	97.6%	100%	100%	99.6%	Xanthinuria, type II, 603592
MOCS1	100%	100%	100%	100%	99.4%	Molybdenum cofactor deficiency A, 252150

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MOCS2	100%	100%	100%	100%	99.9%	Molybdenum cofactor deficiency B1, 252160
MOGS	100%	100%	100%	100%	99.5%	Congenital disorder of glycosylation, type IIb, 606056
MORC2	100%	100%	100%	100%	99.3%	Charcot-Marie-Tooth disease, axonal, type 2Z, 616688;Developmental delay, impaired growth, dysmorphic facies, and axonal neuropathy, 619090
MPC2	100%	100%	100%	99.9%	99.7%	
MPDU1	100%	100%	100%	100%	98.8%	Congenital disorder of glycosylation, type If, 609180
MPI	100%	100%	100%	100%	99.7%	Congenital disorder of glycosylation, type Ib, 602579
MRPL44	100%	100%	100%	100%	99.7%	Combined oxidative phosphorylation deficiency 16, 615395
MSMO1	100%	100%	100%	100%	99.9%	Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834
MTHFD1	94.9%	92%	100%	100%	99.7%	{Neural tube defects, folate-sensitive, susceptibility to}, 601634;Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia, 617780

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MTHFR	100%	100%	100%	100%	99.5%	{Vascular disease, susceptibility to};Homocystinuria due to MTHFR deficiency, 236250;{Thromboembolism, susceptibility to}, 188050;{Schizophrenia, susceptibility to}, 181500;{Neural tube defects, susceptibility to}, 601634
MTM1	100%	100%	99.2%	92.2%	74.7%	Myopathy, centronuclear, X-linked, 310400
MTMR2	100%	100%	100%	99.9%	99.6%	Charcot-Marie-Tooth disease, type 4B1, 601382
MTR	100%	100%	100%	100%	99.7%	{Neural tube defects, folate-sensitive, susceptibility to}, 601634;Homocystinuria-megaloblastic anemia, cblG complementation type, 250940
MTRR	100%	100%	100%	100%	99.7%	Homocystinuria-megaloblastic anemia, cbl E type, 236270;{Neural tube defects, folate-sensitive, susceptibility to}, 601634
MVK	100%	100%	100%	100%	99.8%	Hyper-IgD syndrome, 260920;Porokeratosis 3, multiple types, 175900;Mevalonic aciduria, 610377
NADK2	100%	100%	100%	99.9%	99.2%	2,4-dienoyl-CoA reductase deficiency, 616034
NAGA	90%	88%	100%	100%	99.5%	Schindler disease, type I, 609241;Kanzaki disease, 609242;Schindler disease, type III, 609241

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NAGLU	100%	100%	100%	100%	99.4%	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491;Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920
NAGS	100%	100%	100%	100%	99.2%	N-acetylglutamate synthase deficiency, 237310
NANS	94.8%	94.3%	100%	99.9%	99.7%	Spondyloepimetaphyseal dysplasia, Genevieve type, 610442
NAXD	95.5%	89.7%	100%	100%	99.2%	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 2, 618321
NAXE	97.6%	93.5%	100%	100%	99.5%	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186
NBAS	100%	100%	100%	100%	99.7%	Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800;Infantile liver failure syndrome 2, 616483
NDST1	100%	100%	100%	100%	99.2%	Intellectual developmental disorder, autosomal recessive 46, 616116
NEU1	100%	100%	100%	99.9%	99.5%	Sialidosis, type II, 256550;Sialidosis, type I, 256550
NGLY1	98.4%	97.2%	100%	100%	99.8%	Congenital disorder of deglycosylation 1, 615273

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NMNAT1	100%	99.5%	100%	100%	99.4%	Spondyloepiphyseal dysplasia, sensorineural hearing loss, intellectual developmental disorder, and Leber congenital amaurosis, 619260;Leber congenital amaurosis 9, 608553
NNT	96.3%	96.3%	100%	100%	99.8%	Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency, 614736
NPC1	100%	100%	100%	100%	99.7%	Niemann-Pick disease, type C1, 257220;Niemann-Pick disease, type D, 257220
NPC2	99%	95.2%	100%	100%	99.6%	Niemann-pick disease, type C2, 607625
NPL	91.7%	91.7%	100%	100%	99.7%	
NRDC	96.9%	96.9%	100%	100%	99.8%	
NSD1	100%	100%	100%	100%	99.5%	Sotos syndrome, 117550
NSDHL	95.9%	95.9%	99.3%	93.1%	75.4%	CK syndrome, 300831;CHILD syndrome, 308050
NT5C3A	100%	100%	100%	100%	99.9%	Anemia, congenital, nonspherocytic hemolytic, 8, 266120
NT5E	91%	91%	100%	100%	99.5%	Calcification of joints and arteries, 211800
NUS1	93.9%	93.9%	100%	100%	99.4%	Intellectual developmental disorder, autosomal dominant 55, with seizures, 617831;?Congenital disorder of glycosylation, type 1aa, 617082

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OAT	100%	100%	100%	99.9%	99.3%	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870
OCRL	100%	100%	99.3%	92.4%	74.8%	Dent disease 2, 300555;Lowe syndrome, 309000
ODC1	100%	100%	100%	100%	99.8%	Bachmann-Bupp syndrome, 619075
OGDH	100%	100%	100%	100%	99.5%	Oxoglutarate dehydrogenase deficiency, 203740
OGDHL	100%	100%	100%	100%	99.5%	
OGT	100%	100%	99.2%	92%	73.4%	Intellectual developmental disorder, X-linked 106, 300997
OPA3	100%	100%	100%	100%	99.6%	3-methylglutaconic aciduria, type III, 258501;Optic atrophy 3 with cataract, 165300
OPLAH	100%	100%	100%	100%	99.3%	5-oxoprolinase deficiency, 260005
OSTC	78.3%	78.3%	100%	100%	99.9%	
OTC	100%	100%	99.6%	91.8%	73.3%	Ornithine transcarbamylase deficiency, 311250
OXCT1	90.5%	90.5%	100%	100%	99.7%	Succinyl CoA:3-oxoacid CoA transferase deficiency, 245050
PAH	94.9%	93.7%	100%	100%	99.8%	[Hyperphenylalaninemia, non-PKU mild], 261600;Phenylketonuria, 261600
PAICS	100%	100%	100%	100%	99.8%	?Phosphoribosylaminoimidazole carboxylase deficiency, 619859
PANK2	100%	100%	100%	100%	99.6%	Neurodegeneration with brain iron accumulation 1, 234200

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PAPSS2	100%	100%	100%	100%	99.6%	Brachyolmia 4 with mild epiphyseal and metaphyseal changes, 612847
PC	99.9%	99.2%	100%	100%	99.5%	Pyruvate carboxylase deficiency, 266150
PCBD1	100%	100%	100%	100%	99.5%	Hyperphenylalaninemia, BH4-deficient, D, 264070
PCCA	84.6%	84.4%	100%	100%	99.7%	Propionicacidemia, 606054
PCCB	86.8%	85.3%	100%	100%	99.8%	Propionicacidemia, 606054
PCK1	100%	100%	100%	100%	99.7%	Phosphoenolpyruvate carboxykinase deficiency, cytosolic, 261680
PCK2	100%	100%	100%	100%	99.5%	PEPCK deficiency, mitochondrial, 261650
PCSK9	95%	95%	100%	100%	99.5%	{Low density lipoprotein cholesterol level QTL 1}, 603776;Hypercholesterolemia, familial, 3, 603776
PCYT1A	100%	100%	100%	100%	99.7%	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940;Lipodystrophy, congenital generalized, type 5, 620680
PCYT2	99.9%	97.1%	100%	100%	99.5%	Spastic paraplegia 82, autosomal recessive, 618770
PDSS1	100%	100%	100%	100%	99.7%	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	100%	100%	100%	100%	99.4%	Coenzyme Q10 deficiency, primary, 3, 614652
PEPD	94%	94%	100%	100%	99.6%	Prolidase deficiency, 170100

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PEX1	100%	100%	100%	100%	99.9%	Heimler syndrome 1, 234580;Peroxisome biogenesis disorder 1B (NALD/IRD), 601539;Peroxisome biogenesis disorder 1A (Zellweger), 214100
PEX10	100%	100%	100%	99.6%	98.7%	Peroxisome biogenesis disorder 6A (Zellweger), 614870;Peroxisome biogenesis disorder 6B, 614871
PEX11B	100%	100%	100%	99.9%	99.1%	Peroxisome biogenesis disorder 14B, 614920
PEX12	100%	100%	100%	100%	99.5%	Peroxisome biogenesis disorder 3B, 266510;Peroxisome biogenesis disorder 3A (Zellweger), 614859
PEX13	92.4%	92.4%	100%	100%	99.9%	Peroxisome biogenesis disorder 11A (Zellweger), 614883;Peroxisome biogenesis disorder 11B, 614885
PEX14	100%	100%	100%	100%	99.2%	Peroxisome biogenesis disorder 13A (Zellweger), 614887
PEX16	100%	100%	100%	100%	99.6%	Peroxisome biogenesis disorder 8B, 614877;Peroxisome biogenesis disorder 8A (Zellweger), 614876
PEX19	88.6%	88.6%	100%	100%	99.9%	Peroxisome biogenesis disorder 12A (Zellweger), 614886
PEX2	100%	100%	100%	100%	99.7%	Peroxisome biogenesis disorder 5A (Zellweger), 614866;Peroxisome biogenesis disorder 5B, 614867

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PEX26	91.9%	91.7%	100%	99.9%	99.1%	Peroxisome biogenesis disorder 7B, 614873;Peroxisome biogenesis disorder 7A (Zellweger), 614872
PEX3	100%	100%	100%	100%	99.9%	Peroxisome biogenesis disorder 10A (Zellweger), 614882;?Peroxisome biogenesis disorder 10B, 617370
PEX5	100%	100%	100%	99.8%	99%	Peroxisome biogenesis disorder 2B, 202370;Peroxisome biogenesis disorder 2A (Zellweger), 214110;Rhizomelic chondrodysplasia punctata, type 5, 616716
PEX6	100%	100%	100%	100%	99.1%	Peroxisome biogenesis disorder 4B, 614863;Peroxisome biogenesis disorder 4A (Zellweger), 614862;Heimler syndrome 2, 616617
PEX7	97.9%	97.9%	100%	100%	99.6%	Rhizomelic chondrodysplasia punctata, type 1, 215100;Peroxisome biogenesis disorder 9B, 614879
PFAS	100%	100%	100%	100%	99.3%	
PFKM	100%	100%	100%	100%	99.7%	Glycogen storage disease VII, 232800
PGAM2	100%	100%	100%	99.8%	99.4%	Glycogen storage disease X, 261670
PGAP1	96.7%	96.7%	100%	100%	99.9%	Neurodevelopmental disorder with dysmorphic features, spasticity, and brain abnormalities, 615802

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PGAP2	100%	100%	100%	99.9%	98.9%	Hyperphosphatasia with impaired intellectual development syndrome 3, 614207
PGAP3	100%	100%	100%	100%	99.4%	Hyperphosphatasia with impaired intellectual development syndrome 4, 615716
PGK1	92.7%	92.7%	99.3%	90.9%	71.5%	Phosphoglycerate kinase 1 deficiency, 300653
PGM1	87.5%	87.5%	100%	100%	99.4%	Congenital disorder of glycosylation, type It, 614921
PGM2L1	100%	100%	100%	100%	99.9%	Neurodevelopmental disorder with hypotonia, dysmorphic facies, and skin abnormalities, 620191
PGM3	100%	100%	100%	100%	99.8%	Immunodeficiency 23, 615816
PHGDH	100%	100%	100%	100%	99.6%	Neu-Laxova syndrome 1, 256520;Phosphoglycerate dehydrogenase deficiency, 601815
PHKA1	100%	100%	99.5%	91.9%	72.6%	Muscle glycogenosis, 300559
PHKA2	100%	100%	99.1%	90.1%	71%	Glycogen storage disease, type IXa2, 306000;Glycogen storage disease, type IXa1, 306000
PHKB	100%	100%	100%	100%	99.7%	Phosphorylase kinase deficiency of liver and muscle, autosomal recessive, 261750
PHKG1	97.4%	92.5%	100%	100%	99.3%	
PHKG2	100%	100%	100%	100%	99.1%	Glycogen storage disease IXc, 613027

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PHYH	100%	100%	100%	99.8%	99.1%	Refsum disease, 266500
PI4K2A	86.9%	86.9%	100%	100%	99.1%	Neurodevelopmental disorder with hyperkinetic movements, seizures and structural brain abnormalities, 620732
PI4KA	99.3%	99.2%	100%	99.9%	99%	Spastic paraplegia 84, autosomal recessive, 619621;Gastrointestinal defects and immunodeficiency syndrome 2, 619708;Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis, 616531
PIGA	100%	100%	99.2%	90.8%	73.4%	Paroxysmal nocturnal hemoglobinuria, somatic, 300818;Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868;Neurodevelopmental disorder with epilepsy and hemochromatosis, 301072
PIGB	100%	100%	100%	100%	99.8%	Developmental and epileptic encephalopathy 80, 618580
PIGC	100%	100%	100%	100%	99.8%	Glycosylphosphatidylinositol biosynthesis defect 16, 617816
PIGF	100%	100%	100%	100%	99.8%	Onychodystrophy, osteodystrophy, impaired intellectual development, and seizures syndrome, 619356

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PIGG	100%	100%	100%	100%	99.5%	[Blood group, EMM system], 619812;Neurodevelopmental disorder with or without hypotonia, seizures, and cerebellar atrophy, 616917
PIGH	82.9%	76.2%	100%	100%	99.8%	Glycosylphosphatidylinositol biosynthesis defect 17, 618010
PIGK	88.2%	88.2%	100%	100%	99.9%	Neurodevelopmental disorder with hypotonia and cerebellar atrophy, with or without seizures, 618879
PIGL	100%	100%	100%	100%	99.7%	CHIME syndrome, 280000
PIGM	100%	100%	100%	100%	99.8%	Glycosylphosphatidylinositol deficiency, 610293
PIGN	95.9%	94.4%	100%	100%	99.7%	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	100%	100%	100%	100%	99.8%	Hyperphosphatasia with impaired intellectual development syndrome 2, 614749
PIGP	100%	100%	100%	99.9%	99.4%	Developmental and epileptic encephalopathy 55, 617599
PIGQ	100%	100%	100%	100%	99.6%	Multiple congenital anomalies-hypotonia-seizures syndrome 4, 618548
PIGS	98.4%	95.1%	100%	99.9%	99.1%	Developmental and epileptic encephalopathy 95, 618143

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PIGT	100%	100%	100%	100%	99.5%	?Paroxysmal nocturnal hemoglobinuria 2, 615399;Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398
PIGU	97.1%	97.1%	100%	100%	99.7%	Neurodevelopmental disorder with brain anomalies, seizures, and scoliosis, 618590
PIGV	100%	100%	100%	100%	99.7%	Hyperphosphatasia with impaired intellectual development syndrome 1, 239300
PIGW	100%	100%	100%	100%	99.9%	Glycosylphosphatidylinositol biosynthesis defect 11, 616025
PIGY	100%	100%	100%	99.8%	99.3%	Hyperphosphatasia with impaired intellectual development syndrome 6, 616809

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PIK3CA	100%	100%	100%	100%	99.9%	Hemifacial myohyperplasia, somatic, 606773;CLOVE syndrome, somatic, 612918;Hepatocellular carcinoma, somatic, 114550;Breast cancer, somatic, 114480;Cerebral cavernous malformations 4, somatic, 619538;Ovarian cancer, somatic, 167000;Colorectal cancer, somatic, 114500;Macrodactyly, somatic, 155500;CLAPO syndrome, somatic, 613089;Keratosis, seborrheic, somatic, 182000;Gastric cancer, somatic, 613659;Nonsmall cell lung cancer, somatic, 211980;Nevus, epidermal, somatic mosaic, 162900;Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501;Cowden syndrome 5, 615108
PIK3R1	100%	100%	100%	100%	99.7%	Immunodeficiency 36, 616005;SHORT syndrome, 269880;Agammaglobulinemia 7, autosomal recessive, 615214
PIK3R2	100%	100%	100%	100%	99.2%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387
PIK3R5	100%	100%	100%	100%	98.9%	Ataxia-oculomotor apraxia 3, 615217
PIKFYVE	100%	100%	100%	100%	99.8%	Corneal fleck dystrophy, 121850

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PIP5K1C	90.6%	90.2%	100%	99.9%	99.4%	Lethal congenital contractural syndrome 3, 611369
PKLR	100%	100%	100%	100%	99.4%	Anemia, congenital, nonspherocytic hemolytic, 2, pyruvate kinase deficient, 266200;[Adenosine triphosphate, elevated, of erythrocytes], 102900
PLA2G5	100%	100%	100%	100%	99.3%	[Fleck retina, familial benign], 228980
PLA2G6	100%	100%	100%	100%	99.4%	Parkinson disease 14, autosomal recessive, 612953;Neurodegeneration with brain iron accumulation 2B, 610217;Infantile neuroaxonal dystrophy 1, 256600
PLA2G7	100%	100%	100%	100%	99.6%	Platelet-activating factor acetylhydrolase deficiency, 614278
PLAAT3	65.6%	63.1%	100%	100%	99.7%	Lipodystrophy, familial partial, type 9, 620683
PLCB1	100%	100%	100%	100%	99.6%	Developmental and epileptic encephalopathy 12, 613722
PLCB4	99%	99%	100%	100%	99.8%	Auriculocondylar syndrome 2B, 620458;Auriculocondylar syndrome 2A, 614669
PLCD1	100%	100%	100%	100%	99.6%	Nail disorder, nonsyndromic congenital, 3, (leukonychia), 151600
PLCE1	100%	100%	100%	100%	99.8%	Nephrotic syndrome, type 3, 610725

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PLCG2	96.3%	96.3%	100%	100%	99.3%	Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878;Familial cold autoinflammatory syndrome 3, 614468
PLIN1	100%	100%	100%	99.9%	99.1%	Lipodystrophy, familial partial, type 4, 613877
PLOD1	96.5%	96.5%	100%	100%	99.2%	Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400
PLOD2	100%	100%	100%	99.9%	99.4%	Bruck syndrome 2, 609220
PLOD3	98.5%	96.8%	100%	99.8%	98.9%	BCARD syndrome (lysyl hydroxylase 3 deficiency), 612394
PLPBP	100%	100%	100%	99.8%	98.6%	Epilepsy, early-onset, 1, vitamin B6-dependent, 617290
PMM2	94.6%	94.6%	100%	100%	99.7%	Congenital disorder of glycosylation, type Ia, 212065
PNLIP	100%	100%	100%	100%	99.6%	?Pancreatic lipase deficiency, 614338
PNMT	100%	100%	100%	99.9%	99.3%	
PNP	100%	100%	100%	100%	99.8%	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
PNPLA2	80.4%	80.4%	100%	100%	99.6%	Neutral lipid storage disease with myopathy, 610717
PNPLA6	100%	100%	100%	100%	99.3%	Spastic paraplegia 39, autosomal recessive, 612020;Oliver-McFarlane syndrome, 275400;?Laurence-Moon syndrome, 245800;Boucher-Neuhauser syndrome, 215470

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PNPO	100%	100%	100%	100%	99.6%	Pyridoxamine 5'-phosphate oxidase deficiency, 610090
POFUT1	100%	100%	100%	99.9%	99.6%	Dowling-Degos disease 2, 615327
POGLUT1	99.9%	98%	100%	100%	99.8%	Dowling-Degos disease 4, 615696;Muscular dystrophy, limb-girdle, autosomal recessive 21, 617232
POLD1	100%	100%	100%	99.9%	99%	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381;Immunodeficiency 120, 620836;{Colorectal cancer, susceptibility to, 10}, 612591
POLR3A	100%	100%	100%	100%	99.6%	Wiedemann-Rautenstrauch syndrome, 264090;Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694
POLR3B	95.4%	95.4%	100%	100%	99.7%	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381;Charcot-Marie-Tooth disease, demyelinating, type 1I, 619742

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POMGNT1	100%	100%	100%	100%	99.6%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157;Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 3, 613151;Retinitis pigmentosa 76, 617123;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280
POMGNT2	100%	100%	100%	100%	99.5%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 8, 614830;Muscular dystrophy-dystroglycanopathy (limb-girdle) type C, 8, 618135
POMK	100%	100%	100%	100%	99.8%	?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249
POMT1	100%	99.7%	100%	100%	99.3%	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 impaired intellectual development), type B, 1, 613155

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POMT2	94.9%	94.9%	100%	99.8%	98.4%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150;Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 2, 613156
PPARG	95.8%	95.8%	100%	100%	99.8%	{Diabetes, type 2}, 125853;Insulin resistance, severe, digenic, 125853;Lipodystrophy, familial partial, type 3, 604367;Obesity, severe, 601665
PPCDC	100%	100%	100%	100%	100%	
PPCS	100%	100%	100%	100%	99.3%	Cardiomyopathy, dilated, 2C, 618189
PPFIA3	100%	100%	100%	99.8%	98.5%	Paul-Chao neurodevelopmental syndrome, 621122
PPM1K	100%	100%	100%	100%	99.8%	Maple syrup urine disease, mild variant, 615135
PPOX	99.9%	97.6%	100%	100%	99.3%	Variegate porphyria, childhood-onset, 620483;Variegate porphyria, 176200
PPT1	90.3%	90.3%	100%	100%	99.7%	Ceroid lipofuscinosis, neuronal, 1, 256730
PRKAG2	100%	100%	100%	99.9%	99.6%	Glycogen storage disease of heart, lethal congenital, 261740;Wolff-Parkinson-White syndrome, 194200;Cardiomyopathy, hypertrophic 6, 600858

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PRKCSH	100%	99.6%	100%	99.9%	99.4%	Polycystic liver disease 1, 174050
PRODH	91.9%	91.9%	100%	99.9%	99.5%	{Schizophrenia, susceptibility to, 4}, 600850;Hyperprolinemia, type I, 239500
PRPS1	100%	100%	99%	89.8%	71.8%	Arts syndrome, 301835;Phosphoribosyl pyrophosphate synthetase superactivity, 300661;Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070;Deafness, X-linked 1, 304500;Gout, PRPS-related, 300661
PSAP	99.6%	97.1%	100%	100%	99.4%	Combined SAP deficiency, 611721;Krabbe disease, atypical, 611722;Metachromatic leukodystrophy due to SAP-b deficiency, 249900;Gaucher disease, atypical, 610539;{Parkinson disease 24, autosomal dominant, susceptibility to}, 619491
PSAT1	100%	100%	100%	100%	99.9%	Neu-Laxova syndrome 2, 616038;Phosphoserine aminotransferase deficiency, 610992
PSPH	100%	100%	100%	100%	99.8%	Phosphoserine phosphatase deficiency, 614023

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PTEN	89.6%	89.6%	100%	100%	99.6%	{Glioma susceptibility 2}, 613028;{Meningioma}, 607174;Cowden syndrome 1, 158350;Lhermitte-Duclos disease, 158350;Prostate cancer, somatic, 176807;Macrocephaly/autism syndrome, 605309
PTGIS	100%	100%	100%	100%	99.6%	Hypertension, essential, 145500
PTPN11	89.2%	89.2%	100%	100%	99.8%	Noonan syndrome 1, 163950;LEOPARD syndrome 1, 151100;Metachondromatosis, 156250;Leukemia, juvenile myelomonocytic, somatic, 607785
PTS	100%	100%	100%	100%	99.6%	Hyperphenylalaninemia, BH4-deficient, A, 261640
PUS3	100%	100%	100%	100%	100%	Neurodevelopmental disorder with microcephaly and gray sclerae, 617051
PYCR1	100%	100%	100%	100%	99.8%	Cutis laxa, autosomal recessive, type IIIB, 614438;Cutis laxa, autosomal recessive, type IIB, 612940
PYCR2	100%	100%	100%	100%	99.5%	Leukodystrophy, hypomyelinating, 10, 616420
PYGL	100%	100%	100%	99.9%	99.4%	Glycogen storage disease VI, 232700
PYGM	100%	100%	100%	100%	99.6%	McArdle disease, 232600
QDPR	93.7%	93.6%	100%	100%	99.3%	Hyperphenylalaninemia, BH4-deficient, C, 261630

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RBCK1	100%	100%	100%	99.8%	98.8%	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895
RDH12	100%	100%	100%	100%	99.6%	Leber congenital amaurosis 13, 612712
RDH5	100%	100%	100%	100%	99.5%	Fundus albipunctatus, 136880
RFT1	84.9%	84.9%	100%	100%	99.8%	Congenital disorder of glycosylation, type In, 612015
RINT1	93.6%	93.6%	100%	100%	99.8%	Infantile liver failure syndrome 3, 618641
RPE65	100%	100%	100%	100%	99.8%	Retinitis pigmentosa 20, 613794;Retinitis pigmentosa 87 with choroidal involvement, 618697;Leber congenital amaurosis 2, 204100
RPIA	100%	100%	100%	100%	99.7%	Ribose 5-phosphate isomerase deficiency, 608611
RPN2	96.5%	96.5%	100%	100%	99.8%	
RXYLT1	100%	100%	100%	99.8%	99.3%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041
SAMD9	100%	100%	100%	100%	99.8%	Tumoral calcinosis, familial, normophosphatemic, 610455;Monosomy 7 myelodysplasia and leukemia syndrome 2, 619041;MIRAGE syndrome, 617053

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SAMD9L	100%	100%	100%	100%	99.9%	Ataxia-pancytopenia syndrome, 159550;?Spinocerebellar ataxia 49, 619806;Monosomy 7 myelodysplasia and leukemia syndrome 1, 252270
SARDH	84.7%	84.7%	100%	100%	99.5%	[Sarcosinemia], 268900
SAT1	100%	100%	98.9%	89.4%	69.8%	
SC5D	100%	100%	100%	100%	99.9%	Lathosterolosis, 607330
SCARB2	100%	100%	100%	100%	99.7%	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCP2	95.5%	95.5%	100%	100%	99.7%	?Leukoencephalopathy with dystonia and motor neuropathy, 613724
SCYL1	100%	100%	100%	99.9%	99.1%	Spinocerebellar ataxia, autosomal recessive 21, 616719
SEC23B	100%	100%	100%	100%	99.7%	?Cowden syndrome 7, 616858;Dyserythropoietic anemia, congenital, type II, 224100
SEC63	92.9%	92.9%	100%	100%	99.9%	Polycystic liver disease 2, 617004
SELENBP1	93.1%	93.1%	100%	100%	99.4%	Extraoral halitosis due to MTO deficiency, 618148
SEPHS1	91.3%	86.6%	100%	100%	99.8%	Ververi-Brady syndrome 2, 621325
SEPSECS	99.4%	96.3%	100%	100%	99.5%	Pontocerebellar hypoplasia type 2D, 613811
SERAC1	100%	100%	100%	100%	99.9%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SGMS1	100%	100%	100%	100%	99.5%	

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SGSH	100%	100%	100%	100%	99.5%	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900
SHMT2	100%	100%	100%	100%	99.7%	Neurodevelopmental disorder with cardiomyopathy, spasticity, and brain abnormalities, 619121
SI	100%	100%	100%	99.7%	99.1%	Sucrase-isomaltase deficiency, congenital, 222900
SLC10A7	92.8%	92.8%	100%	100%	100%	Short stature, amelogenesis imperfecta, and skeletal dysplasia with scoliosis, 618363
SLC12A1	96.3%	96.3%	100%	100%	99.8%	Bartter syndrome, type 1, 601678
SLC13A3	100%	100%	100%	100%	99.6%	Leukoencephalopathy, acute reversible, with increased urinary alpha-ketoglutarate, 618384
SLC16A1	100%	100%	100%	100%	99.9%	Hyperinsulinemic hypoglycemia, familial, 7, 610021;Erythrocyte lactate transporter defect, 245340;Monocarboxylate transporter 1 deficiency, 616095
SLC17A5	92.7%	92.7%	100%	100%	99.8%	Salla disease, 604369;Sialic acid storage disorder, infantile, 269920
SLC18A2	99.6%	96.7%	100%	100%	99.8%	Parkinsonism-dystonia, infantile, 2, 618049
SLC1A1	100%	100%	100%	99.9%	99.4%	Dicarboxylic aminoaciduria, 222730;{?Schizophrenia susceptibility 18}, 615232

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SLC1A4	100%	100%	100%	99.9%	99.3%	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657
SLC22A12	100%	100%	100%	99.9%	98.7%	Hypouricemia, renal, 220150
SLC22A5	97.1%	97.1%	100%	100%	99.5%	Carnitine deficiency, systemic primary, 212140
SLC25A1	100%	100%	100%	99.9%	98.8%	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182;Myasthenic syndrome, congenital, 23, presynaptic, 618197
SLC25A13	91.7%	91.7%	100%	100%	99.6%	Citrullinemia, type II, neonatal-onset, 605814;Citrullinemia, adult-onset type II, 603471
SLC25A15	100%	100%	100%	100%	99.8%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A19	100%	100%	100%	100%	99.8%	Microcephaly, Amish type, 607196;Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710
SLC25A20	100%	100%	100%	100%	99.4%	Carnitine-acylcarnitine translocase deficiency, 212138
SLC25A21	100%	100%	100%	100%	99.9%	?Mitochondrial DNA depletion syndrome 18, 618811
SLC25A32	100%	100%	100%	100%	99.7%	?Exercise intolerance, riboflavin-responsive, 616839
SLC25A36	95%	95%	100%	100%	99.4%	Hyperinsulinemic hypoglycemia, familial, 8, 620211

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SLC25A38	90.6%	90.6%	100%	100%	99.4%	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950
SLC25A42	100%	100%	100%	100%	99.2%	Metabolic crises, recurrent, with variable encephalomyopathic features and neurologic regression, 618416
SLC26A2	100%	100%	100%	100%	99.8%	Epiphyseal dysplasia, multiple, 4, 226900;De la Chapelle dysplasia, 256050;Diastrophic dysplasia, 222600;Diastrophic dysplasia, broad bone-platyspondylic variant, 222600;Achondrogenesis IB, 600972;Atelosteogenesis, type II, 256050
SLC27A3	100%	100%	100%	99.9%	99%	
SLC28A1	100%	100%	100%	100%	99.6%	[Uridine-cytidineuria], 618477
SLC29A3	100%	100%	100%	100%	99.5%	Histiocytosis-lymphadenopathy plus syndrome, 602782
SLC2A1	100%	100%	100%	100%	99.3%	Dystonia 9, 601042;GLUT1 deficiency syndrome 1, infantile onset, severe, 606777;Stomatin-deficient cryohydrocytosis with neurologic defects, 608885;{Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847;GLUT1 deficiency syndrome 2, childhood onset, 612126
SLC2A2	100%	100%	100%	100%	99.8%	Fanconi-Bickel syndrome, 227810;{Diabetes mellitus, noninsulin-dependent}, 125853

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SLC2A9	100%	100%	100%	100%	99.7%	{Uric acid concentration, serum, QTL 2}, 612076;Hypouricemia, renal, 2, 612076
SLC30A10	100%	100%	100%	100%	99.3%	Hypermanganesemia with dystonia 1, 613280
SLC30A2	99.9%	97.9%	100%	100%	99.6%	Zinc deficiency, transient neonatal, 608118
SLC30A9	100%	100%	100%	100%	99.9%	Birk-Landau-Perez syndrome, 617595
SLC33A1	92.6%	92.6%	100%	100%	99.7%	Spastic paraplegia 42, autosomal dominant, 612539;Huppke-Brendel syndrome, 614482
SLC35A1	94.9%	94.9%	100%	100%	100%	Congenital disorder of glycosylation, type IIf, 603585
SLC35A2	100%	100%	98.9%	87.9%	68.3%	Congenital disorder of glycosylation, type IIm, 300896
SLC35A3	91%	87.6%	100%	100%	99.8%	Arthrogryposis, impaired intellectual development, and seizures, 615553
SLC35B2	100%	100%	100%	100%	99.2%	Leukodystrophy, hypomyelinating, 26, with chondrodysplasia, 620269
SLC35C1	100%	100%	100%	100%	98.9%	Congenital disorder of glycosylation, type IIc, 266265
SLC35D1	100%	100%	100%	100%	99.8%	Schneckenbecken dysplasia, 269250
SLC36A2	100%	100%	100%	100%	99.8%	[Iminoglycinuria], 242600;[Hyperglycinuria], 138500

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SLC37A4	100%	100%	100%	100%	99.8%	Glycogen storage disease Ib, 232220;Congenital disorder of glycosylation, type IIw, 619525;Glycogen storage disease Ic, 232240
SLC38A3	96.3%	96.3%	100%	100%	99.4%	Developmental and epileptic encephalopathy 102, 619881
SLC39A14	94%	94%	100%	100%	99.5%	?Hyperostosis cranalis interna, 144755;Hypermanganesemia with dystonia 2, 617013
SLC39A4	100%	100%	100%	100%	99.5%	Acrodermatitis enteropathica, 201100
SLC39A8	77.5%	77.4%	100%	100%	100%	Congenital disorder of glycosylation, type IIn, 616721
SLC3A1	96.2%	96.2%	100%	100%	99.8%	Cystinuria, 220100
SLC44A1	97.5%	97.5%	100%	100%	99.8%	Neurodegeneration, childhood-onset, with ataxia, tremor, optic atrophy, and cognitive decline, 618868
SLC45A1	100%	100%	100%	100%	99.4%	Intellectual developmental disorder with neuropsychiatric features, 617532
SLC46A1	100%	100%	99.9%	99.7%	99%	Folate malabsorption, hereditary, 229050
SLC52A1	100%	100%	100%	100%	99.9%	Riboflavin deficiency, 615026
SLC52A2	100%	100%	100%	99.9%	99.7%	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	100%	100%	100%	100%	99.4%	?Fazio-Londe disease, 211500;Brown-Vialetto-Van Laere syndrome 1, 211530

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SLC5A1	100%	100%	100%	100%	99.4%	Glucose/galactose malabsorption, 606824
SLC5A2	100%	100%	100%	99.9%	99.4%	Renal glucosuria, 233100
SLC6A19	100%	100%	100%	99.9%	99%	Hartnup disorder, 234500
SLC6A5	100%	100%	100%	100%	99.6%	Hyperekplexia 3, 614618
SLC6A6	100%	100%	100%	100%	99.4%	Hypotaurinemic retinal degeneration and cardiomyopathy, 145350
SLC6A8	100%	99.9%	98.4%	88.1%	71.2%	Cerebral creatine deficiency syndrome 1, 300352
SLC6A9	100%	100%	100%	100%	99.2%	Glycine encephalopathy with normal serum glycine, 617301;{Scoliosis, isolated, susceptibility to, 6}, 621428
SLC7A7	100%	100%	100%	100%	98.8%	Lysinuric protein intolerance, 222700
SLC7A9	100%	100%	100%	100%	99.5%	Cystinuria, 220100
SLC9A7	100%	100%	99%	89%	70.1%	Intellectual developmental disorder, X-linked 108, 301024
SLCO1B1	100%	100%	100%	99.9%	99.6%	Hyperbilirubinemia, Rotor type, digenic, 237450
SLCO1B3	100%	100%	100%	100%	99.9%	Hyperbilirubinemia, Rotor type, digenic, 237450
SMPD1	100%	100%	100%	100%	99.6%	Niemann-Pick disease, type B, 607616;Niemann-Pick disease, type A, 257200

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SMS	94.4%	94.2%	99.1%	91.9%	73.3%	Intellectual developmental disorder, X-linked syndromic, Snyder-Robinson type, 309583
SNX14	95%	95%	100%	100%	99.8%	Spinocerebellar ataxia, autosomal recessive 20, 616354
SOD1	100%	100%	100%	100%	99.7%	Spastic tetraplegia and axial hypotonia, progressive, 618598;Amyotrophic lateral sclerosis 1, 105400
SOD2	100%	100%	100%	100%	99.8%	{Microvascular complications of diabetes 6}, 612634
SPR	100%	100%	100%	100%	99.8%	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SPTLC1	84.4%	83.8%	100%	100%	99.7%	Amyotrophic lateral sclerosis 27, juvenile, 620285;Neuropathy, hereditary sensory and autonomic, type IA, 162400
SPTLC2	89%	89%	100%	100%	99.7%	Neuropathy, hereditary sensory and autonomic, type IC, 613640
SPTSSA	100%	100%	100%	100%	99.9%	Spastic paraplegia 90A, autosomal dominant, 620416;?Spastic paraplegia 90B, autosomal recessive, 620417
SQOR	100%	100%	100%	100%	99.7%	Sulfide:quinone oxidoreductase deficiency, 619221
SRD5A2	87.5%	87.5%	100%	100%	99.3%	Pseudovaginal perineoscrotal hypospadias, 264600

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SRD5A3	100%	100%	100%	100%	99.9%	Kahrizi syndrome, 612713;Congenital disorder of glycosylation, type Iq, 612379
SSR3	100%	100%	100%	100%	99.4%	
SSR4	100%	100%	98.9%	87.6%	65.5%	Congenital disorder of glycosylation, type Iy, 300934
ST3GAL3	89.5%	88.2%	100%	100%	99.5%	Developmental and epileptic encephalopathy 15, 615006;Intellectual developmental disorder, autosomal recessive 12, 611090
ST3GAL5	98.4%	98.4%	100%	100%	99.6%	Salt and pepper developmental regression syndrome, 609056
STAR	100%	100%	100%	100%	99.4%	Lipoid adrenal hyperplasia, 201710
STS	97.1%	97.1%	98.9%	90.5%	73.5%	Ichthyosis, X-linked, 308100
STT3A	99.9%	98.8%	100%	100%	99.8%	Congenital disorder of glycosylation, type Iw, autosomal dominant, 619714;Congenital disorder of glycosylation, type Iw, autosomal recessive, 615596
STT3B	100%	100%	100%	100%	99.5%	Congenital disorder of glycosylation, type Ix, 615597
STX5	100%	100%	100%	100%	99.7%	?Congenital disorder of glycosylation, type IIaa, 620454
SUCLA2	96.3%	96.3%	100%	100%	99.9%	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073

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SUCLG1	100%	100%	100%	100%	99.8%	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400
SUCLG2	100%	100%	100%	100%	99.9%	
SUGCT	100%	100%	100%	100%	99.7%	Glutaric aciduria III, 231690
SUMF1	100%	100%	100%	100%	99.9%	Multiple sulfatase deficiency, 272200
SUOX	100%	100%	100%	100%	99.5%	Sulfite oxidase deficiency, 272300
TAFAZZIN	100%	100%	98.6%	89.7%	70.1%	Barth syndrome, 302060
TALDO1	89.1%	89.1%	100%	100%	99.7%	Transaldolase deficiency, 606003
TANGO2	100%	100%	100%	100%	99.6%	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878
TAT	100%	100%	100%	100%	99.8%	Tyrosinemia, type II, 276600
TBXAS1	100%	100%	100%	100%	99.8%	Ghosal hematodiaphyseal syndrome, 231095
TCIRG1	100%	100%	100%	100%	99.7%	Osteopetrosis, autosomal recessive 1, 259700
TCN2	91.8%	87.7%	100%	100%	99.9%	Transcobalamin II deficiency, 275350
TECR	87.3%	87.3%	100%	100%	98.9%	Intellectual developmental disorder, autosomal recessive 14, 614020
TH	100%	99.8%	100%	100%	99.2%	Segawa syndrome, recessive, 605407

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TIMM50	100%	100%	100%	100%	99.6%	3-methylglutaconic aciduria, type IX, 617698
TK2	100%	100%	100%	100%	99.9%	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560;?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3, 617069
TKFC	100%	100%	100%	99.9%	99.4%	Triokinase and FMN cyclase deficiency syndrome, 618805
TKT	89%	89%	100%	100%	99.5%	Short stature, developmental delay, and congenital heart defects, 617044
TMEM106B	100%	100%	100%	100%	99.9%	Leukodystrophy, hypomyelinating, 16, 617964
TMEM165	88.9%	88.9%	100%	100%	99.7%	Congenital disorder of glycosylation, type IIk, 614727
TMEM260	94.8%	94.8%	100%	100%	99.7%	Structural heart defects and renal anomalies syndrome, 617478
TMEM70	100%	100%	100%	100%	99.3%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052
TMLHE	94.8%	94.8%	99.6%	94.8%	79.1%	{Autism, susceptibility to, X-linked 6}, 300872
TMTC3	100%	100%	100%	100%	99.7%	Lissencephaly 8, 617255
TNIK	98.6%	98.6%	100%	100%	99.6%	Intellectual developmental disorder, autosomal recessive 54, 617028
TPI1	100%	100%	100%	100%	99.6%	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512

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TPK1	93.3%	93.3%	100%	100%	99.9%	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458
TPMT	100%	100%	100%	100%	99.7%	{Thiopurines, poor metabolism of, 1}, 610460
TPP1	100%	100%	100%	100%	99.3%	Ceroid lipofuscinosis, neuronal, 2, 204500;Spinocerebellar ataxia, autosomal recessive 7, 609270
TRAK1	100%	100%	100%	100%	99.7%	Developmental and epileptic encephalopathy 68, 618201
TRAPPC11	100%	100%	100%	100%	99.8%	Muscular dystrophy, limb-girdle, autosomal recessive 18, 615356
TRAPPC2L	100%	100%	100%	100%	99.6%	Encephalopathy, progressive, early-onset, with episodic rhabdomyolysis, 618331
TRAPPC9	100%	100%	100%	100%	99.5%	Intellectual developmental disorder, autosomal recessive 13, 613192
TREH	100%	100%	100%	100%	99.7%	Trehalase deficiency, 612119
TREX1	100%	100%	100%	100%	99.5%	Vasculopathy, retinal, with cerebral leukoencephalopathy and systemic manifestations, 192315;Aicardi-Goutieres syndrome 1, dominant and recessive, 225750;{Systemic lupus erythematosus, susceptibility to}, 152700;Chilblain lupus, 610448

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TRIP11	100%	100%	100%	100%	99.8%	Odontochondrodysplasia 1, 184260;Achondrogenesis, type IA, 200600
TUSC3	93.3%	93.3%	100%	100%	99.7%	Intellectual developmental disorder, autosomal recessive 7, 611093
TXNIP	100%	100%	100%	100%	99.7%	
TYMP	100%	100%	100%	100%	99.4%	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041
TYMS	89.8%	85.5%	100%	100%	99.7%	Dyskeratosis congenita, digenic, 620040
TYR	100%	100%	100%	100%	99.7%	[Skin/hair/eye pigmentation 3, light/dark/freckling skin], 601800;[Skin/hair/eye pigmentation 3, blue/green eyes], 601800;{Melanoma, cutaneous malignant, susceptibility to, 8}, 601800;Albinism, oculocutaneous, type IB, 606952;Albinism, oculocutaneous, type IA, 203100
TYRP1	100%	100%	100%	100%	99.7%	[Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)], 612271;Albinism, oculocutaneous, type III, 203290
UFM1	100%	100%	100%	100%	99.8%	Leukodystrophy, hypomyelinating, 14, 617899
UGDH	97%	94%	100%	100%	99.7%	Developmental and epileptic encephalopathy 84, 618792

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UGGT1	100%	100%	100%	100%	99.8%	Congenital disorder of glycosylation, type IICC, 621381
UGP2	91.8%	90.6%	100%	100%	99.8%	Developmental and epileptic encephalopathy 83, 618744
UGT1A1	100%	100%	100%	100%	99.6%	Crigler-Najjar syndrome, type I, 218800;[Bilirubin, serum level of, QTL1], 601816;Hyperbilirubinemia, familial transient neonatal, 237900;Crigler-Najjar syndrome, type II, 606785;[Gilbert syndrome], 143500
UMPS	90.6%	90.6%	100%	100%	99.6%	Orotic aciduria, 258900
UPB1	94.3%	91.8%	100%	100%	99.5%	Beta-ureidopropionase deficiency, 613161
UROC1	96.6%	96.6%	100%	100%	99.6%	?Urocanase deficiency, 276880
UROD	100%	100%	100%	100%	99.6%	Porphyria, hepatoerythropoietic, 176100;Porphyria cutanea tarda, 176100
UROS	95.3%	95.3%	100%	100%	99.9%	Porphyria, congenital erythropoietic, 263700
VMA12	100%	100%	100%	99.9%	99.2%	Congenital disorder of glycosylation, type IIp, 616829
VMA21	100%	100%	98.5%	86.5%	67.4%	Myopathy, X-linked, with excessive autophagy, 310440
VMA22	100%	98.5%	100%	99.9%	97.9%	Congenital disorder of glycosylation, type IIo, 616828
VPS13B	100%	100%	100%	100%	99.7%	Cohen syndrome, 216550
VPS33A	90.3%	90.2%	100%	100%	99.7%	Mucopolysaccharidosis-plus syndrome, 617303

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VPS51	100%	100%	100%	100%	99%	Pontocerebellar hypoplasia, type 13, 618606
VPS53	83.1%	81.5%	100%	99.9%	99.4%	Pontocerebellar hypoplasia, type 2E, 615851
XDH	100%	100%	100%	100%	99.6%	Xanthinuria, type I, 278300
XYLT1	100%	100%	100%	99.9%	99.3%	Desbuquois dysplasia 2, 615777;{Pseudoxanthoma elasticum, modifier of severity of}, 264800
XYLT2	99.9%	98.8%	100%	99.9%	99.5%	{Pseudoxanthoma elasticum, modifier of severity of}, 264800;Spondyloocular syndrome, 605822
YME1L1	96.7%	96.7%	100%	100%	99.7%	?Optic atrophy 11, 617302
ZBTB11	100%	100%	100%	100%	99.8%	Intellectual developmental disorder, autosomal recessive 69, 618383
ZMPSTE24	100%	100%	100%	100%	100%	Mandibuloacral dysplasia with type B lipodystrophy, 608612;Restrictive dermopathy 1, 275210
ZNF143	94.2%	94.2%	100%	100%	99.6%	

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 X2 covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.
TWIST X2 covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.
srWGS covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WGS mapped against GRCh38.
srWGS covered 15x describes the percentage of a gene's coding sequence that is covered at least 15x when analyzed by WGS mapped against GRCh38.
srWGS covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WGS mapped against GRCh38.
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 : November 25th, 2024.
This list is accurate for panel version DG 5.0.0
Ad 1. Blank field signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors