# METABOLIC DISORDERS PANEL DG-4.0.0 (749 GENES)

| *Gene* | *Twist X2 covered >10x* | *Twist X2 covered >20x* | *WGS covered >10x* | *WGS covered >20x* | *Associated Phenotype description and OMIM disease ID* |
|---|---|---|---|---|---|
| AASS | 100.0% | 100.0% | 100.0% | 98.3% | Hyperlysinemia, 238700 |
| ABAT | 100.0% | 100.0% | 100.0% | 98.7% | GABA-transaminase deficiency, 613163 |
| ABCC8 | 100.0% | 100.0% | 100.0% | 99.4% | Diabetes mellitus, permanent neonatal 3, with or without neurologic features, 618857;Diabetes mellitus, transient neonatal 2, 610374;Diabetes mellitus, noninsulin-dependent, 125853;Hypoglycemia of infancy, leucine-sensitive, 240800;Hyperinsulinemic hypoglycemia, familial, 1, 256450 |
| ABCD1 | 100.0% | 99.6% | 98.9% | 76.9% | Adrenoleukodystrophy, 300100;Adrenomyeloneuropathy, adult, 300100 |
| ABCD2 | 100.0% | 100.0% | 100.0% | 98.4% | |
| ABCD3 | 100.0% | 100.0% | 100.0% | 97.2% | ?Bile acid synthesis defect, congenital, 5, 616278 |

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| ABCD4 | 100.0% | 100.0% | 100.0% | 98.8% | Methylmalonic aciduria and homocystinuria, cblJ type, 614857 |
| ABCG5 | 100.0% | 100.0% | 100.0% | 98.3% | Sitosterolemia 2, 618666 |
| ABCG8 | 100.0% | 100.0% | 100.0% | 99.2% | Sitosterolemia 1, 210250;{Gallbladder disease 4}, 611465 |
| ABHD12 | 100.0% | 100.0% | 99.9% | 97.3% | Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674 |
| ABHD5 | 100.0% | 100.0% | 100.0% | 99.0% | Chanarin-Dorfman syndrome, 275630 |
| ACACA | 100.0% | 100.0% | 100.0% | 99.0% | Acetyl-CoA carboxylase deficiency, 613933 |
| ACAD8 | 100.0% | 100.0% | 100.0% | 99.1% | Isobutyryl-CoA dehydrogenase deficiency, 611283 |
| ACAD9 | 100.0% | 100.0% | 100.0% | 99.6% | Mitochondrial complex I deficiency, nuclear type 20, 611126 |
| ACADM | 95.3% | 94.0% | 100.0% | 97.3% | Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450 |
| ACADS | 100.0% | 100.0% | 100.0% | 99.5% | Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470 |
| ACADSB | 100.0% | 100.0% | 100.0% | 98.9% | 2-methylbutyrylglycinuria, 610006 |
| ACADVL | 100.0% | 100.0% | 99.9% | 96.4% | VLCAD deficiency, 201475 |

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| ACAT1 | 100.0% | 100.0% | 99.8% | 95.7% | Alpha-methylacetoacetic aciduria, 203750 |
| ACAT2 | 100.0% | 100.0% | 100.0% | 98.6% | ?ACAT2 deficiency, 614055 |
| ACBD5 | 85.7% | 85.6% | 99.9% | 96.1% | Retinal dystrophy with leukodystrophy, 618863 |
| ACBD6 | 100.0% | 100.0% | 100.0% | 97.5% | Neurodevelopmental disorder with progressive movement abnormalities, 620785 |
| ACO2 | 92.4% | 89.8% | 100.0% | 99.3% | Optic atrophy 9, 616289;Infantile cerebellar-retinal degeneration, 614559 |
| ACOX1 | 100.0% | 100.0% | 100.0% | 99.1% | Mitchell syndrome, 618960;Peroxisomal acyl-CoA oxidase deficiency, 264470 |
| ACOX2 | 100.0% | 100.0% | 100.0% | 99.3% | Bile acid synthesis defect, congenital, 6, 617308 |
| ACSF3 | 100.0% | 100.0% | 100.0% | 98.8% | Combined malonic and methylmalonic aciduria, 614265 |
| ACSL4 | 100.0% | 100.0% | 97.8% | 72.3% | Intellectual developmental disorder, X-linked 63, 300387 |
| ACY1 | 100.0% | 100.0% | 100.0% | 99.4% | Aminoacylase 1 deficiency, 609924 |

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| ADA | 85.0% | 84.2% | 100.0% | 99.4% | Adenosine deaminase deficiency, partial, 102700;Severe combined immunodeficiency due to ADA deficiency, 102700 |
| ADCK5 | 100.0% | 100.0% | 100.0% | 99.0% | |
| ADCY5 | 97.4% | 97.3% | 100.0% | 97.4% | Dyskinesia with orofacial involvement, autosomal dominant, 606703;Neurodevelopmental disorder with hyperkinetic movements and dyskinesia, 619651;Dyskinesia with orofacial involvement, autosomal recessive, 619647 |
| ADK | 90.9% | 90.9% | 100.0% | 98.6% | Hypermethioninemia due to adenosine kinase deficiency, 614300 |
| ADSL | 100.0% | 100.0% | 100.0% | 99.0% | Adenylosuccinase deficiency, 103050 |
| AGA | 100.0% | 100.0% | 100.0% | 98.3% | Aspartylglucosaminuria, 208400 |
| AGK | 91.7% | 91.7% | 100.0% | 98.9% | Cataract 38, autosomal recessive, 614691;Sengers syndrome, 212350 |
| AGL | 100.0% | 100.0% | 100.0% | 98.1% | Glycogen storage disease IIIa, 232400;Glycogen storage disease IIIb, 232400 |
| AGPAT2 | 100.0% | 100.0% | 100.0% | 97.9% | Lipodystrophy, congenital generalized, type 1, 608594 |

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| AGPS | 97.3% | 97.3% | 100.0% | 96.8% | Rhizomelic chondrodysplasia punctata, type 3, 600121 |
| AGXT | 100.0% | 100.0% | 100.0% | 99.6% | Hyperoxaluria, primary, type 1, 259900 |
| AHCY | 100.0% | 100.0% | 100.0% | 99.4% | Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752 |
| AK1 | 100.0% | 100.0% | 100.0% | 99.6% | Hemolytic anemia due to adenylate kinase deficiency, 612631 |
| AK2 | 100.0% | 100.0% | 100.0% | 99.6% | Reticular dysgenesis, 267500 |
| AKR1C1 | 100.0% | 100.0% | 100.0% | 98.7% | |
| AKR1D1 | 100.0% | 100.0% | 100.0% | 98.2% | Bile acid synthesis defect, congenital, 2, 235555 |
| ALAD | 100.0% | 100.0% | 100.0% | 99.6% | Porphyria, acute hepatic, 612740;{Lead poisoning, susceptibility to}, 612740 |
| ALAS2 | 100.0% | 99.8% | 98.3% | 72.9% | Anemia, sideroblastic, 1, 300751;Protoporphyria, erythropoietic, X-linked, 300752 |

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| ALDH18A1 | 100.0% | 100.0% | 100.0% | 99.5% | Spastic paraplegia 9A, autosomal dominant, 601162;Cutis laxa, autosomal recessive, type IIIA, 219150;Spastic paraplegia 9B, autosomal recessive, 616586;Cutis laxa, autosomal dominant 3, 616603 |
| ALDH1A3 | 100.0% | 100.0% | 100.0% | 97.6% | Microphthalmia, isolated 8, 615113 |
| ALDH2 | 100.0% | 100.0% | 100.0% | 98.6% | Alcohol sensitivity, acute, 610251;{Hangover, susceptibility to}, 610251;{Esophageal cancer, alcohol-related, susceptibility to}, ;{Sublingual nitroglycerin, susceptibility to poor response to}, |
| ALDH3A2 | 93.5% | 93.5% | 100.0% | 98.4% | Sjogren-Larsson syndrome, 270200 |
| ALDH4A1 | 100.0% | 100.0% | 100.0% | 98.6% | Hyperprolinemia, type II, 239510 |
| ALDH5A1 | 100.0% | 100.0% | 100.0% | 97.8% | Succinic semialdehyde dehydrogenase deficiency, 271980 |
| ALDH6A1 | 100.0% | 100.0% | 99.9% | 97.1% | Methylmalonate semialdehyde dehydrogenase deficiency, 614105 |

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| ALDH7A1 | 100.0% | 100.0% | 100.0% | 99.0% | Epilepsy, early-onset, 4, vitamin B6-dependent, 266100 |
| ALDOA | 100.0% | 100.0% | 100.0% | 99.7% | Glycogen storage disease XII, 611881 |
| ALDOB | 100.0% | 100.0% | 100.0% | 99.4% | Fructose intolerance, hereditary, 229600 |
| ALG1 | 100.0% | 100.0% | 100.0% | 99.4% | Congenital disorder of glycosylation, type Ik, 608540 |
| ALG10 | 100.0% | 100.0% | 100.0% | 98.1% | |
| ALG11 | 91.0% | 91.0% | 100.0% | 98.4% | Congenital disorder of glycosylation, type Ip, 613661 |
| ALG12 | 100.0% | 100.0% | 100.0% | 99.8% | Congenital disorder of glycosylation, type Ig, 607143 |
| ALG13 | 99.7% | 99.0% | 97.0% | 70.4% | Developmental and epileptic encephalopathy 36, 300884 |
| ALG14 | 100.0% | 100.0% | 100.0% | 98.2% | 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 | 100.0% | 100.0% | 100.0% | 99.4% | Congenital disorder of glycosylation, type Ii, 607906;Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228 |
| ALG3 | 100.0% | 100.0% | 100.0% | 97.8% | Congenital disorder of glycosylation, type Id, 601110 |
| ALG6 | 100.0% | 100.0% | 99.9% | 96.4% | Congenital disorder of glycosylation, type Ic, 603147 |
| ALG8 | 78.1% | 77.5% | 100.0% | 97.9% | Congenital disorder of glycosylation, type Ih, 608104;Polycystic liver disease 3 with or without kidney cysts, 617874 |
| ALG9 | 100.0% | 100.0% | 100.0% | 98.6% | Gillessen-Kaesbach-Nishimura syndrome, 263210;Congenital disorder of glycosylation, type Il, 608776 |
| ALOX12B | 100.0% | 100.0% | 100.0% | 98.5% | Ichthyosis, congenital, autosomal recessive 2, 242100 |
| ALPL | 100.0% | 100.0% | 100.0% | 99.5% | Odontohypophosphatasia, 146300;Hypophosphatasia, infantile, 241500;Hypophosphatasia, childhood, 241510;Hypophosphatasia, adult, 146300 |

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| AMACR | 100.0% | 100.0% | 100.0% | 97.1% | Alpha-methylacyl-CoA racemase deficiency, 614307;Bile acid synthesis defect, congenital, 4, 214950 |
| AMN | 100.0% | 100.0% | 100.0% | 97.9% | Imerslund-Grasbeck syndrome 2, 618882 |
| AMPD1 | 100.0% | 100.0% | 100.0% | 98.3% | Myopathy due to myoadenylate deaminase deficiency, 615511 |
| AMPD3 | 100.0% | 100.0% | 100.0% | 99.2% | [AMP deaminase deficiency, erythrocytic], 612874 |
| AMT | 100.0% | 100.0% | 100.0% | 99.5% | Glycine encephalopathy 2, 620398 |
| AP1S1 | 100.0% | 100.0% | 100.0% | 98.3% | MEDNIK syndrome, 609313 |
| AP3B2 | 100.0% | 100.0% | 100.0% | 98.8% | Developmental and epileptic encephalopathy 48, 617276 |
| APOA5 | 100.0% | 100.0% | 100.0% | 99.4% | Hyperchylomicronemia, late-onset, 144650;{Hypertriglyceridemia, susceptibility to}, 145750 |
| APOC2 | 100.0% | 100.0% | 100.0% | 97.6% | Hyperlipoproteinemia, type Ib, 207750 |
| APRT | 100.0% | 100.0% | 100.0% | 99.0% | Adenine phosphoribosyltransferase deficiency, 614723 |
| ARG1 | 93.0% | 93.0% | 100.0% | 98.7% | Argininemia, 207800 |
| ARSA | 100.0% | 100.0% | 100.0% | 99.4% | Metachromatic leukodystrophy, 250100 |

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| ARSB | 100.0% | 100.0% | 100.0% | 97.4% | Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200 |
| ASAH1 | 100.0% | 100.0% | 100.0% | 97.8% | Spinal muscular atrophy with progressive myoclonic epilepsy, 159950;Farber lipogranulomatosis, 228000 |
| ASL | 100.0% | 100.0% | 100.0% | 99.3% | Argininosuccinic aciduria, 207900 |
| ASNS | 100.0% | 100.0% | 100.0% | 98.2% | Asparagine synthetase deficiency, 615574 |
| ASPA | 100.0% | 100.0% | 100.0% | 98.2% | Canavan disease, 271900 |
| ASS1 | 100.0% | 100.0% | 100.0% | 99.7% | Citrullinemia, 215700 |
| ATIC | 100.0% | 100.0% | 100.0% | 97.8% | AICA-ribosiduria due to ATIC deficiency, 608688 |
| ATP1A1 | 100.0% | 100.0% | 100.0% | 99.1% | Hypomagnesemia, seizures, and impaired intellectual development 2, 618314;Charcot-Marie-Tooth disease, axonal, type 2DD, 618036 |
| ATP6AP1 | 100.0% | 99.7% | 98.3% | 70.6% | Immunodeficiency 47, 300972 |
| ATP6AP2 | 100.0% | 100.0% | 97.4% | 70.6% | Intellectual developmental disorder, X-linked syndromic, Hedera type, 300423;?Parkinsonism with spasticity, X-linked, 300911;Congenital disorder of glycosylation, type IIr, 301045 |

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| ATP6V0A2 | 100.0% | 100.0% | 100.0% | 97.2% | Wrinkly skin syndrome, 278250;Cutis laxa, autosomal recessive, type IIA, 219200 |
| ATP6V1A | 100.0% | 100.0% | 100.0% | 97.9% | Cutis laxa, autosomal recessive, type IID, 617403;Developmental and epileptic encephalopathy 93, 618012 |
| ATP6V1E1 | 100.0% | 100.0% | 100.0% | 98.1% | Cutis laxa, autosomal recessive, type IIC, 617402 |
| ATP7A | 94.9% | 94.5% | 98.1% | 71.7% | Occipital horn syndrome, 304150;Neuronopathy, distal hereditary motor, X-linked, 300489;Menkes disease, 309400 |
| ATP7B | 100.0% | 100.0% | 100.0% | 99.3% | Wilson disease, 277900 |
| ATP8B1 | 100.0% | 100.0% | 100.0% | 97.4% | Cholestasis, progressive familial intrahepatic 1, 211600;Cholestasis, intrahepatic, of pregnancy, 1, 147480;Cholestasis, benign recurrent intrahepatic, 243300 |
| AUH | 100.0% | 100.0% | 100.0% | 97.1% | 3-methylglutaconic aciduria, type I, 250950 |
| B3GALNT1 | 100.0% | 100.0% | 100.0% | 97.4% | [Blood group, P1PK system, P(k) phenotype], 111400;[Blood group, globoside system], 615021 |

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| B3GALNT2 | 92.4% | 92.4% | 100.0% | 97.7% | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 11, 615181 |
| B3GALT6 | 99.9% | 98.0% | 100.0% | 94.8% | Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349;Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640;Al-Gazali syndrome, 609465 |
| B3GAT3 | 94.5% | 93.8% | 100.0% | 98.7% | Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600 |
| B3GLCT | 100.0% | 100.0% | 100.0% | 98.0% | Peters-plus syndrome, 261540 |
| B4GALT1 | 100.0% | 100.0% | 100.0% | 98.6% | Combined low LDL and fibrinogen, 620364;Congenital disorder of glycosylation, type IId, 607091 |
| B4GALT7 | 100.0% | 100.0% | 100.0% | 99.0% | Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070 |
| B4GAT1 | 100.0% | 100.0% | 100.0% | 98.7% | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287 |

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| BAAT | 100.0% | 100.0% | 100.0% | 99.0% | Bile acid conjugation defect 1, 619232 |
| BCAT1 | 100.0% | 100.0% | 100.0% | 99.0% | |
| BCAT2 | 100.0% | 100.0% | 100.0% | 99.5% | ?Hypervalinemia or hyperleucine-isoleucinemia, 618850 |
| BCKDHA | 100.0% | 100.0% | 100.0% | 99.4% | Maple syrup urine disease, type Ia, 248600 |
| BCKDHB | 100.0% | 99.8% | 100.0% | 97.4% | Maple syrup urine disease, type Ib, 620698 |
| BCKDK | 100.0% | 100.0% | 100.0% | 99.3% | Branched-chain keto acid dehydrogenase kinase deficiency, 614923 |
| BCO1 | 100.0% | 100.0% | 100.0% | 99.6% | ?Hypercarotenemia and vitamin A deficiency, autosomal dominant, 115300 |
| BLVRA | 100.0% | 99.9% | 100.0% | 98.8% | Hyperbiliverdinemia, 614156 |
| BMP2 | 100.0% | 100.0% | 100.0% | 98.1% | Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies 1, 617877;Brachydactyly, type A2, 112600;{HFE hemochromatosis, modifier of}, 235200 |
| BPGM | 100.0% | 100.0% | 100.0% | 98.9% | Erythrocytosis, familial, 8, 222800 |

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| BPNT2 | 100.0% | 100.0% | 100.0% | 98.5% | Chondrodysplasia with joint dislocations, GPAPP type, 614078 |
| BSCL2 | 100.0% | 100.0% | 100.0% | 99.3% | 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% | 100.0% | 99.5% | Biotinidase deficiency, 253260 |
| C1GALT1C1 | 100.0% | 100.0% | 98.4% | 72.0% | Hemolytic uremic syndrome, atypical, 8, with rhizomelic short stature, 301110;Tn polyagglutination syndrome, somatic, 300622 |
| C2orf69 | 100.0% | 100.0% | 99.9% | 96.5% | Combined oxidative phosphorylation deficiency 53, 619423 |
| CA5A | 100.0% | 100.0% | 100.0% | 98.0% | Hyperammonemia due to carbonic anhydrase VA deficiency, 615751 |
| CAD | 100.0% | 100.0% | 100.0% | 99.4% | Developmental and epileptic encephalopathy 50, 616457 |
| CANT1 | 100.0% | 100.0% | 100.0% | 99.5% | Desbuquois dysplasia 1, 251450;Epiphyseal dysplasia, multiple, 7, 617719 |

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| CAT | 100.0% | 100.0% | 100.0% | 98.3% | Acatalasemia, 614097 |
| CAV1 | 74.6% | 74.6% | 100.0% | 98.7% | Lipodystrophy, congenital generalized, type 3, 612526;Pulmonary hypertension, primary, 3, 615343;Lipodystrophy, familial partial, type 7, 606721 |
| CAVIN1 | 100.0% | 100.0% | 100.0% | 98.1% | Lipodystrophy, congenital generalized, type 4, 613327 |
| CBLIF | 100.0% | 100.0% | 100.0% | 99.1% | Intrinsic factor deficiency, 261000 |
| CBS | 100.0% | 100.0% | 100.0% | 99.5% | Thrombosis, hyperhomocysteinemic, 236200;Homocystinuria, B6-responsive and nonresponsive types, 236200 |
| CCDC115 | 100.0% | 100.0% | 100.0% | 96.7% | Congenital disorder of glycosylation, type IIo, 616828 |
| CD320 | 100.0% | 100.0% | 100.0% | 99.7% | Methylmalonic aciduria, transient, due to transcobalamin receptor defect, 613646 |
| CEL | 100.0% | 100.0% | 99.3% | 92.2% | Maturity-onset diabetes of the young, type VIII, 609812 |
| CERKL | 98.8% | 98.4% | 100.0% | 97.7% | Retinitis pigmentosa 26, 608380 |

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| CERS3 | 100.0% | 100.0% | 100.0% | 98.1% | Ichthyosis, congenital, autosomal recessive 9, 615023 |
| CFTR | 100.0% | 100.0% | 100.0% | 98.6% | Cystic fibrosis, 219700;Congenital bilateral absence of vas deferens, 277180;{Pancreatitis, hereditary}, 167800;{Bronchiectasis with or without elevated sweat chloride 1, modifier of}, 211400;Sweat chloride elevation without CF, ;{Hypertrypsinemia, neonatal}, |
| CHIT1 | 100.0% | 100.0% | 100.0% | 99.0% | [Chitotriosidase deficiency], 614122 |
| CHKB | 100.0% | 100.0% | 100.0% | 98.7% | Muscular dystrophy, congenital, megaconial type, 602541 |
| CHST14 | 100.0% | 100.0% | 100.0% | 91.9% | Ehlers-Danlos syndrome, musculocontractural type 1, 601776 |
| CHST3 | 100.0% | 100.0% | 100.0% | 99.7% | Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095 |
| CHST6 | 100.0% | 100.0% | 100.0% | 99.9% | Macular corneal dystrophy, 217800 |
| CHSY1 | 99.9% | 99.7% | 100.0% | 97.5% | Temtamy preaxial brachydactyly syndrome, 605282 |
| CIAO1 | 100.0% | 100.0% | 100.0% | 99.5% | |

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| CIDEC | 100.0% | 100.0% | 100.0% | 98.7% | ?Lipodystrophy, familial partial, type 5, 615238 |
| CLCN7 | 100.0% | 100.0% | 100.0% | 99.4% | Hypopigmentation, organomegaly, and delayed myelination and development, 618541;Osteopetrosis, autosomal recessive 4, 611490;Osteopetrosis, autosomal dominant 2, 166600 |
| CLN3 | 93.2% | 93.1% | 100.0% | 98.5% | Ceroid lipofuscinosis, neuronal, 3, 204200 |
| CLN5 | 83.1% | 83.0% | 100.0% | 96.8% | Ceroid lipofuscinosis, neuronal, 5, 256731 |
| CLN6 | 100.0% | 100.0% | 100.0% | 97.6% | Ceroid lipofuscinosis, neuronal, 6B (Kufs type), 204300;Ceroid lipofuscinosis, neuronal, 6A, 601780 |
| CLN8 | 100.0% | 100.0% | 100.0% | 99.6% | Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003;Ceroid lipofuscinosis, neuronal, 8, 600143 |

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| CLPB | 100.0% | 100.0% | 99.9% | 98.3% | Neutropenia, severe congenital, 9, autosomal dominant, 619813;3-methylglutaconic aciduria, type VIIB, autosomal recessive, 616271;3-methylglutaconic aciduria, type VIIA, autosomal dominant, 619835 |
| CMAS | 100.0% | 100.0% | 100.0% | 98.2% | |
| COG1 | 100.0% | 100.0% | 100.0% | 97.4% | Congenital disorder of glycosylation, type IIg, 611209 |
| COG2 | 100.0% | 100.0% | 100.0% | 98.6% | ?Congenital disorder of glycosylation, type IIq, 617395 |
| COG3 | 100.0% | 100.0% | 100.0% | 98.2% | Congenital disorder of glycosylation, type IIbb, 620546 |
| COG4 | 100.0% | 100.0% | 100.0% | 98.6% | Congenital disorder of glycosylation, type IIj, 613489;Saul-Wilson syndrome, 618150 |
| COG5 | 100.0% | 100.0% | 100.0% | 98.3% | Congenital disorder of glycosylation, type IIi, 613612 |
| COG6 | 100.0% | 100.0% | 100.0% | 98.2% | Shaheen syndrome, 615328;Congenital disorder of glycosylation, type III, 614576 |

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| COG7 | 100.0% | 100.0% | 100.0% | 98.0% | Congenital disorder of glycosylation, type IIe, 608779 |
| COG8 | 100.0% | 100.0% | 99.9% | 97.4% | Congenital disorder of glycosylation, type IIh, 611182 |
| COMT | 92.1% | 91.9% | 100.0% | 99.7% | {Schizophrenia, susceptibility to}, 181500;{Panic disorder, susceptibility to}, 167870 |
| COQ2 | 96.3% | 96.3% | 100.0% | 98.5% | {Multiple system atrophy, susceptibility to}, 146500;Coenzyme Q10 deficiency, primary, 1, 607426 |
| COQ4 | 100.0% | 100.0% | 100.0% | 99.6% | Coenzyme Q10 deficiency, primary, 7, 616276;Spastic ataxia 10, autosomal recessive, 620666 |
| COQ5 | 100.0% | 100.0% | 100.0% | 97.4% | ?Coenzyme Q10 deficiency, primary, 9, 619028 |
| COQ6 | 100.0% | 100.0% | 99.9% | 98.4% | Coenzyme Q10 deficiency, primary, 6, 614650 |
| COQ7 | 100.0% | 100.0% | 100.0% | 98.8% | Coenzyme Q10 deficiency, primary, 8, 616733;Neuronopathy, distal hereditary motor, autosomal recessive 9, 620402 |
| COQ8A | 100.0% | 100.0% | 100.0% | 99.7% | Coenzyme Q10 deficiency, primary, 4, 612016 |

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| COQ8B | 100.0% | 100.0% | 100.0% | 99.0% | Nephrotic syndrome, type 9, 615573 |
| COQ9 | 100.0% | 100.0% | 100.0% | 98.8% | Coenzyme Q10 deficiency, primary, 5, 614654 |
| CP | 100.0% | 100.0% | 100.0% | 98.7% | Aceruloplasminemia, 604290 |
| CPOX | 100.0% | 100.0% | 100.0% | 97.3% | Coproporphyria, 121300;Harderoporphyria, 618892 |
| CPS1 | 100.0% | 100.0% | 100.0% | 98.5% | Carbamoylphosphate synthetase I deficiency, 237300;{Pulmonary hypertension, neonatal, susceptibility to}, 615371 |
| CPT1A | 100.0% | 100.0% | 100.0% | 98.5% | CPT deficiency, hepatic, type IA, 255120 |
| CPT2 | 100.0% | 100.0% | 100.0% | 98.7% | {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 | 100.0% | 100.0% | 100.0% | 99.3% | ?Neurodegeneration with brain iron accumulation 8, 617917 |
| CRLS1 | 100.0% | 100.0% | 100.0% | 95.1% | Combined oxidative phosphorylation deficiency 57, 620167 |

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| CRPPA | 100.0% | 100.0% | 100.0% | 98.5% | Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643 |
| CTH | 100.0% | 100.0% | 100.0% | 98.6% | Cystathioninuria, 219500 |
| CTNS | 100.0% | 100.0% | 99.8% | 97.9% | Cystinosis, nephropathic, 219800;Cystinosis, ocular nonnephropathic, 219750;Cystinosis, late-onset juvenile or adolescent nephropathic, 219900;Cystinosis, atypical nephropathic, 219800 |
| CTSA | 100.0% | 99.9% | 100.0% | 98.7% | Galactosialidosis, 256540 |
| CTSC | 94.7% | 94.2% | 100.0% | 98.3% | Periodontitis 1, juvenile, 170650;Haim-Munk syndrome, 245010;Papillon-Lefevre syndrome, 245000 |
| CTSD | 100.0% | 100.0% | 100.0% | 99.5% | Ceroid lipofuscinosis, neuronal, 10, 610127 |
| CTSK | 100.0% | 100.0% | 100.0% | 98.6% | Pycnodysostosis, 265800 |
| CUBN | 100.0% | 100.0% | 100.0% | 99.2% | [Proteinuria, chronic benign], 618884;Imerslund-Grasbeck syndrome 1, 261100 |
| CYB561 | 100.0% | 100.0% | 100.0% | 97.3% | Orthostatic hypotension 2, 618182 |

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| CYB5R3 | 95.5% | 92.0% | 100.0% | 97.8% | Methemoglobinemia, type I, 250800;Methemoglobinemia, type II, 250800 |
| CYP11A1 | 100.0% | 100.0% | 100.0% | 99.3% | Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743 |
| CYP11B1 | 100.0% | 100.0% | 100.0% | 99.6% | Aldosteronism, glucocorticoid-remediable, 103900;Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010 |
| CYP11B2 | 100.0% | 100.0% | 100.0% | 98.8% | Hypoaldosteronism, congenital, due to CMO I deficiency, 203400;Hypoaldosteronism, congenital, due to CMO II deficiency, 610600;Aldosterone to renin ratio raised, ;{Low renin hypertension, susceptibility to}, |
| CYP17A1 | 100.0% | 100.0% | 100.0% | 99.2% | 17,20-lyase deficiency, isolated, 202110;17-alpha-hydroxylase/17,20-lyase deficiency, 202110 |
| CYP19A1 | 100.0% | 99.9% | 100.0% | 98.8% | Aromatase deficiency, 613546;Aromatase excess syndrome, 139300 |

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| CYP1B1 | 100.0% | 100.0% | 100.0% | 98.8% | Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300;Anterior segment dysgenesis 6, multiple subtypes, 617315 |
| CYP21A2 | 100.0% | 99.9% | 100.0% | 99.3% | Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910;Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910 |
| CYP27A1 | 100.0% | 100.0% | 100.0% | 99.4% | Cerebrotendinous xanthomatosis, 213700 |
| CYP27B1 | 100.0% | 100.0% | 100.0% | 99.1% | Vitamin D-dependent rickets, type I, 264700 |
| CYP2R1 | 100.0% | 100.0% | 100.0% | 96.7% | Rickets due to defect in vitamin D 25-hydroxylation deficiency, 600081 |
| CYP2U1 | 100.0% | 100.0% | 100.0% | 96.7% | Spastic paraplegia 56, autosomal recessive, 615030 |
| CYP7B1 | 100.0% | 100.0% | 100.0% | 98.8% | Spastic paraplegia 5A, autosomal recessive, 270800;Bile acid synthesis defect, congenital, 3, 613812 |
| D2HGDH | 100.0% | 100.0% | 100.0% | 99.2% | D-2-hydroxyglutaric aciduria, 600721 |
| DAO | 100.0% | 100.0% | 100.0% | 99.2% | |

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| DBH | 100.0% | 100.0% | 100.0% | 99.5% | Orthostatic hypotension 1, due to DBH deficiency, 223360 |
| DBT | 100.0% | 100.0% | 100.0% | 98.4% | Maple syrup urine disease, type II, 620699 |
| DCXR | 100.0% | 100.0% | 100.0% | 99.7% | [Pentosuria], 260800 |
| DDC | 100.0% | 100.0% | 100.0% | 98.7% | Aromatic L-amino acid decarboxylase deficiency, 608643 |
| DDHD1 | 100.0% | 100.0% | 100.0% | 97.6% | Spastic paraplegia 28, autosomal recessive, 609340 |
| DDOST | 100.0% | 100.0% | 100.0% | 98.8% | Congenital disorder of glycosylation, type Ir, 614507 |
| DEGS1 | 100.0% | 100.0% | 100.0% | 99.1% | Leukodystrophy, hypomyelinating, 18, 618404 |
| DGAT1 | 100.0% | 100.0% | 100.0% | 99.0% | Diarrhea 7, protein-losing enteropathy type, 615863 |
| DGKE | 100.0% | 100.0% | 100.0% | 98.7% | {Hemolytic uremic syndrome, atypical, susceptibility to, 7}, 615008;Nephrotic syndrome, type 7, 615008 |

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| DGUOK | 100.0% | 100.0% | 100.0% | 98.6% | 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 | 100.0% | 100.0% | 100.0% | 99.2% | Desmosterolosis, 602398 |
| DHCR7 | 96.2% | 96.2% | 100.0% | 99.7% | Smith-Lemli-Opitz syndrome, 270400 |
| DHDDS | 73.8% | 73.7% | 100.0% | 98.8% | Developmental delay and seizures with or without movement abnormalities, 617836;?Congenital disorder of glycosylation, type 1bb, 613861;Retinitis pigmentosa 59, 613861 |
| DHFR | 100.0% | 100.0% | 100.0% | 98.0% | Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839 |
| DHODH | 100.0% | 100.0% | 100.0% | 98.9% | Miller syndrome, 263750 |
| DHTKD1 | 100.0% | 100.0% | 100.0% | 98.0% | ?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025;Alpha-aminoadipic and alpha-ketoadipic aciduria, 204750 |

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| DLD | 100.0% | 100.0% | 100.0% | 98.7% | Dihydrolipoamide dehydrogenase deficiency, 246900 |
| DMGDH | 100.0% | 100.0% | 100.0% | 98.5% | Dimethylglycine dehydrogenase deficiency, 605850 |
| DNAJC12 | 100.0% | 100.0% | 100.0% | 97.5% | Hyperphenylalaninemia, mild, non-BH4-deficient, 617384 |
| DNAJC19 | 100.0% | 100.0% | 100.0% | 98.3% | 3-methylglutaconic aciduria, type V, 610198 |
| DNM1L | 100.0% | 100.0% | 100.0% | 98.6% | Optic atrophy 5, 610708;Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388 |
| DNM2 | 100.0% | 100.0% | 100.0% | 98.3% | Centronuclear myopathy 1, 160150;Charcot-Marie-Tooth disease, axonal type 2M, 606482;Charcot-Marie-Tooth disease, dominant intermediate B, 606482;Lethal congenital contracture syndrome 5, 615368 |
| DNMT1 | 99.9% | 99.0% | 100.0% | 99.5% | Neuropathy, hereditary sensory, type IE, 614116;Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121 |

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| DNMT3B | 100.0% | 100.0% | 100.0% | 99.2% | Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860;Facioscapulohumeral muscular dystrophy 4, digenic, 619478 |
| DOLK | 100.0% | 100.0% | 100.0% | 98.1% | Congenital disorder of glycosylation, type Im, 610768 |
| DPAGT1 | 100.0% | 100.0% | 100.0% | 99.3% | Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750;Congenital disorder of glycosylation, type Ij, 608093 |
| DPM1 | 99.2% | 96.7% | 100.0% | 98.2% | Congenital disorder of glycosylation, type Ie, 608799 |
| DPM2 | 100.0% | 100.0% | 100.0% | 99.3% | Congenital disorder of glycosylation, type Iu, 615042 |
| DPM3 | 100.0% | 100.0% | 100.0% | 94.8% | ?Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 15, 618992;Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 15, 612937 |

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| DPYD | 99.8% | 99.6% | 100.0% | 98.6% | Dihydropyrimidine dehydrogenase deficiency, 274270;5-fluorouracil toxicity, 274270 |
| DPYS | 100.0% | 100.0% | 100.0% | 98.3% | Dihydropyrimidinuria, 222748 |
| DTYMK | 100.0% | 100.0% | 100.0% | 99.1% | Neurodegeneration, childhood-onset, with progressive microcephaly, 619847 |
| EBP | 100.0% | 100.0% | 98.7% | 72.8% | MEND syndrome, 300960;Chondrodysplasia punctata, X-linked dominant, 302960 |
| ECHS1 | 100.0% | 100.0% | 100.0% | 96.6% | Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277 |
| EDEM3 | 100.0% | 100.0% | 100.0% | 98.3% | Congenital disorder of glycosylation, type IIv, 619493 |
| ELOVL1 | 100.0% | 100.0% | 100.0% | 99.5% | Ichthyotic keratoderma, spasticity, hypomyelination, and dysmorphic facies, 618527 |
| ELOVL4 | 100.0% | 100.0% | 99.9% | 97.6% | Spinocerebellar ataxia 34, 133190;Stargardt disease 3, 600110;Ichthyosis, spastic quadriplegia, and impaired intellectual development, 614457 |
| ENO3 | 100.0% | 100.0% | 100.0% | 99.3% | Glycogen storage disease XIII, 612932 |

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| EOGT | 98.1% | 94.0% | 100.0% | 99.0% | Adams-Oliver syndrome 4, 615297 |
| EPG5 | 100.0% | 100.0% | 100.0% | 98.4% | Vici syndrome, 242840 |
| EPHX1 | 100.0% | 100.0% | 100.0% | 99.2% | |
| EPHX2 | 100.0% | 100.0% | 100.0% | 98.3% | {Hypercholesterolemia, familial, due to LDLR defect, modifier of}, 143890 |
| ETFA | 82.5% | 82.4% | 99.9% | 96.4% | Glutaric acidemia IIA, 231680 |
| ETFB | 100.0% | 100.0% | 100.0% | 99.6% | Glutaric acidemia IIB, 231680 |
| ETFDH | 93.6% | 92.0% | 100.0% | 98.9% | Glutaric acidemia IIC, 231680 |
| ETHE1 | 100.0% | 100.0% | 100.0% | 97.9% | Ethylmalonic encephalopathy, 602473 |
| EXT1 | 100.0% | 100.0% | 100.0% | 98.8% | Exostoses, multiple, type 1, 133700;Chondrosarcoma, 215300 |
| EXT2 | 100.0% | 100.0% | 100.0% | 99.4% | Seizures, scoliosis, and macrocephaly syndrome, 616682;Exostoses, multiple, type 2, 133701 |
| EYA1 | 100.0% | 100.0% | 100.0% | 99.1% | Branchiootic syndrome 1, 602588;Branchiootorenal syndrome 1, with or without cataracts, 113650;Anterior segment anomalies with or without cataract, 602588;?Otofaciocervical syndrome, 166780 |

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| FA2H | 100.0% | 100.0% | 100.0% | 98.8% | Spastic paraplegia 35, autosomal recessive, 612319 |
| FAH | 100.0% | 100.0% | 100.0% | 98.6% | Tyrosinemia, type I, 276700 |
| FAR1 | 100.0% | 100.0% | 100.0% | 98.9% | Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154;Cataracts, spastic paraparesis, and speech delay, 619338 |
| FBN1 | 100.0% | 100.0% | 100.0% | 99.1% | 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 | 100.0% | 100.0% | 100.0% | 99.0% | Fructose-1,6-bisphosphatase deficiency, 229700 |
| FBP2 | 100.0% | 100.0% | 100.0% | 99.5% | ?Leukodystrophy, childhood-onset, remitting, 619864 |
| FCSK | 100.0% | 100.0% | 100.0% | 99.5% | Congenital disorder of glycosylation with defective fucosylation 2, 618324 |
| FDFT1 | 100.0% | 100.0% | 100.0% | 97.7% | Squalene synthase deficiency, 618156 |

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| FECH | 100.0% | 100.0% | 100.0% | 99.1% | Protoporphyria, erythropoietic, 1, 177000 |
| FH | 100.0% | 100.0% | 100.0% | 98.5% | Leiomyomatosis and renal cell cancer, 150800;Fumarase deficiency, 606812 |
| FKRP | 100.0% | 100.0% | 100.0% | 99.9% | 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 | 100.0% | 100.0% | 100.0% | 98.0% | 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 | 100.0% | 100.0% | 100.0% | 99.4% | Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency, 255100 |
| FMO3 | 100.0% | 100.0% | 100.0% | 98.8% | Trimethylaminuria, 602079 |
| FOLR1 | 100.0% | 100.0% | 100.0% | 99.8% | Neurodegeneration due to cerebral folate transport deficiency, 613068 |
| FTCD | 100.0% | 100.0% | 99.9% | 97.7% | Glutamate formiminotransferase deficiency, 229100 |
| FUCA1 | 100.0% | 100.0% | 100.0% | 98.6% | Fucosidosis, 230000 |
| FUT2 | 100.0% | 100.0% | 100.0% | 99.8% | {Vitamin B12 plasma level QTL1}, 612542;[Bombay phenotype, digenic], 616754;{Norwalk virus infection, resistance to}, |

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| FUT6 | 100.0% | 100.0% | 100.0% | 99.1% | [Fucosyltransferase 6 deficiency], 613852 |
| FUT8 | 100.0% | 99.8% | 100.0% | 99.3% | Congenital disorder of glycosylation with defective fucosylation 1, 618005 |
| G6PC1 | 100.0% | 100.0% | 100.0% | 99.4% | Glycogen storage disease Ia, 232200 |
| G6PC3 | 96.7% | 96.7% | 100.0% | 99.3% | Dursun syndrome, 612541;Neutropenia, severe congenital 4, autosomal recessive, 612541 |
| G6PD | 86.3% | 86.1% | 98.6% | 73.9% | Hemolytic anemia, G6PD deficient (favism), 300908;{Resistance to malaria due to G6PD deficiency}, 611162 |
| GAA | 100.0% | 100.0% | 100.0% | 99.5% | Glycogen storage disease II, 232300 |
| GAD1 | 100.0% | 100.0% | 100.0% | 98.4% | Developmental and epileptic encephalopathy 89, 619124 |
| GALC | 100.0% | 100.0% | 100.0% | 98.5% | Krabbe disease, 245200 |
| GALE | 100.0% | 100.0% | 100.0% | 99.3% | Thrombocytopenia 13, syndromic, 620776;Galactose epimerase deficiency, 230350 |
| GALK1 | 100.0% | 100.0% | 100.0% | 99.5% | Galactokinase deficiency with cataracts, 230200 |
| GALM | 100.0% | 100.0% | 100.0% | 98.2% | Galactosemia IV, 618881 |

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| GALNS | 100.0% | 100.0% | 100.0% | 98.6% | Mucopolysaccharidosis IVA, 253000 |
| GALNT2 | 100.0% | 100.0% | 100.0% | 97.1% | Congenital disorder of glycosylation, type IIt, 618885 |
| GALNT3 | 100.0% | 100.0% | 100.0% | 97.9% | Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900 |
| GALT | 100.0% | 100.0% | 100.0% | 99.2% | Galactosemia, 230400 |
| GAMT | 100.0% | 100.0% | 100.0% | 97.5% | Cerebral creatine deficiency syndrome 2, 612736 |
| GANAB | 100.0% | 100.0% | 100.0% | 99.3% | Polycystic kidney disease 3, 600666 |
| GATM | 100.0% | 100.0% | 100.0% | 97.8% | Cerebral creatine deficiency syndrome 3, 612718;Fanconi renotubular syndrome 1, 134600 |
| GBA1 | 100.0% | 100.0% | 100.0% | 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 |

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| GBA2 | 100.0% | 100.0% | 100.0% | 99.2% | Spastic paraplegia 46, autosomal recessive, 614409 |
| GBE1 | 100.0% | 99.9% | 100.0% | 98.4% | Glycogen storage disease IV, 232500;Polyglucosan body disease, adult form, 263570 |
| GCDH | 100.0% | 100.0% | 100.0% | 99.0% | Glutaricaciduria, type I, 231670 |
| GCH1 | 100.0% | 100.0% | 99.9% | 98.2% | Dystonia, DOPA-responsive, 128230;Hyperphenylalaninemia, BH4-deficient, B, 233910 |
| GCK | 100.0% | 100.0% | 100.0% | 99.6% | MODY, type II, 125851;Diabetes mellitus, permanent neonatal 1, 606176;Hyperinsulinemic hypoglycemia, familial, 3, 602485;Diabetes mellitus, noninsulin-dependent, late onset, 125853 |
| GCLC | 100.0% | 100.0% | 100.0% | 98.3% | {Myocardial infarction, susceptibility to}, 608446;Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency, 230450 |
| GCLM | 100.0% | 100.0% | 100.0% | 96.4% | {Myocardial infarction, susceptibility to}, 608446 |

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| GCSH | 100.0% | 100.0% | 100.0% | 98.1% | Multiple mitochondrial dysfunctions syndrome 7, 620423 |
| GFPT1 | 100.0% | 100.0% | 100.0% | 98.9% | Myasthenia, congenital, 12, with tubular aggregates, 610542 |
| GGPS1 | 100.0% | 100.0% | 100.0% | 98.7% | Muscular dystrophy, congenital hearing loss, and ovarian insufficiency syndrome, 619518 |
| GK | 100.0% | 100.0% | 97.2% | 69.3% | Glycerol kinase deficiency, 307030 |
| GLA | 91.4% | 91.4% | 98.4% | 73.6% | Fabry disease, cardiac variant, 301500;Fabry disease, 301500 |
| GLB1 | 100.0% | 100.0% | 100.0% | 98.9% | GM1-gangliosidosis, type I, 230500;GM1-gangliosidosis, type III, 230650;Mucopolysaccharidosis type IVB (Morquio), 253010;GM1-gangliosidosis, type II, 230600 |
| GLDC | 100.0% | 100.0% | 100.0% | 98.6% | Glycine encephalopathy1, 605899 |
| GLRA1 | 100.0% | 100.0% | 100.0% | 99.3% | Hyperekplexia 1, 149400 |
| GLRX5 | 100.0% | 100.0% | 100.0% | 97.1% | Anemia, sideroblastic, 3, pyridoxine-refractory, 616860;Spasticity, childhood-onset, with hyperglycinemia, 616859 |

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| GLS | 100.0% | 100.0% | 100.0% | 97.9% | Global developmental delay, progressive ataxia, and elevated glutamine, 618412;?Infantile cataract, skin abnormalities, glutamate excess, and impaired intellectual development, 618339;Developmental and epileptic encephalopathy 71, 618328 |
| GLUD1 | 100.0% | 100.0% | 100.0% | 94.8% | Hyperinsulinism-hyperammonemia syndrome, 606762 |
| GLUL | 100.0% | 100.0% | 100.0% | 98.9% | Glutamine deficiency, congenital, 610015;Developmental and epileptic encephalopathy 116, 620806 |
| GLYCTK | 100.0% | 100.0% | 100.0% | 99.8% | D-glyceric aciduria, 220120 |
| GM2A | 100.0% | 100.0% | 100.0% | 99.0% | GM2-gangliosidosis, AB variant, 272750 |
| GMPPA | 100.0% | 100.0% | 100.0% | 99.5% | Alacrima, achalasia, and impaired intellectual development syndrome, 615510 |

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| GMPPB | 100.0% | 100.0% | 100.0% | 99.4% | 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 | 100.0% | 100.0% | 100.0% | 98.8% | |
| GNE | 100.0% | 100.0% | 100.0% | 99.3% | Sialuria, 269921;Thrombocytopenia 12 with or without myopathy, 620757;Nonaka myopathy, 605820 |
| GNMT | 100.0% | 100.0% | 100.0% | 97.9% | Glycine N-methyltransferase deficiency, 606664 |
| GNPAT | 100.0% | 100.0% | 100.0% | 98.0% | Rhizomelic chondrodysplasia punctata, type 2, 222765 |
| GNPTAB | 100.0% | 100.0% | 100.0% | 98.5% | Mucolipidosis III alpha/beta, 252600;Mucolipidosis II alpha/beta, 252500 |
| GNPTG | 100.0% | 100.0% | 100.0% | 96.2% | Mucolipidosis III gamma, 252605 |

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| GNS | 100.0% | 100.0% | 100.0% | 99.1% | Mucopolysaccharidosis type IIID, 252940 |
| GOT1 | 100.0% | 100.0% | 100.0% | 99.0% | Aspartate aminotransferase, serum level of, QTL1, 614419 |
| GOT2 | 100.0% | 100.0% | 100.0% | 99.2% | Developmental and epileptic encephalopathy 82, 618721 |
| GPD1 | 100.0% | 100.0% | 100.0% | 99.2% | Hypertriglyceridemia, transient infantile, 614480 |
| GPD1L | 100.0% | 100.0% | 100.0% | 97.7% | Brugada syndrome 2, 611777 |
| GPHN | 100.0% | 99.9% | 100.0% | 98.1% | Molybdenum cofactor deficiency C, 615501 |
| GPI | 100.0% | 100.0% | 100.0% | 98.8% | Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency, 613470 |
| GPIHBP1 | 100.0% | 100.0% | 100.0% | 99.0% | Hyperlipoproteinemia, type 1D, 615947 |
| GPT2 | 100.0% | 100.0% | 100.0% | 98.3% | Neurodevelopmental disorder with microcephaly and spastic paraplegia, 616281 |
| GPX1 | 100.0% | 100.0% | 100.0% | 97.3% | Hemolytic anemia due to glutathione peroxidase deficiency, 614164 |
| GRHPR | 100.0% | 100.0% | 100.0% | 98.9% | Hyperoxaluria, primary, type II, 260000 |

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| GSS | 100.0% | 100.0% | 100.0% | 98.9% | Hemolytic anemia due to glutathione synthetase deficiency, 231900;Glutathione synthetase deficiency, 266130 |
| GUSB | 100.0% | 100.0% | 100.0% | 99.0% | Mucopolysaccharidosis VII, 253220 |
| GYG1 | 100.0% | 100.0% | 100.0% | 98.7% | ?Glycogen storage disease XV, 613507;Polyglucosan body myopathy 2, 616199 |
| GYS1 | 100.0% | 100.0% | 100.0% | 99.0% | Glycogen storage disease 0, muscle, 611556 |
| GYS2 | 100.0% | 100.0% | 100.0% | 98.6% | Glycogen storage disease 0, liver, 240600 |
| H6PD | 100.0% | 100.0% | 100.0% | 99.4% | Cortisone reductase deficiency 1, 604931 |
| HADH | 100.0% | 100.0% | 100.0% | 98.1% | Hyperinsulinemic hypoglycemia, familial, 4, 609975;3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 |
| HADHA | 100.0% | 100.0% | 100.0% | 98.8% | HELLP syndrome, maternal, of pregnancy, 609016;LCHAD deficiency, 609016;Mitochondrial trifunctional protein deficiency 1, 609015;Fatty liver, acute, of pregnancy, 609016 |
| HADHB | 100.0% | 100.0% | 100.0% | 99.0% | Mitochondrial trifunctional protein deficiency 2, 620300 |

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| HAGH | 100.0% | 100.0% | 100.0% | 98.4% | [Glyoxalase II deficiency], 614033 |
| HCFC1 | 100.0% | 99.9% | 98.4% | 75.7% | Methylmalonic aciduria and homocysteinemia, cblX type, 309541 |
| HEXA | 100.0% | 100.0% | 100.0% | 99.1% | [Hex A pseudodeficiency], 272800;GM2-gangliosidosis, several forms, 272800;Tay-Sachs disease, 272800 |
| HEXB | 100.0% | 100.0% | 100.0% | 97.5% | Sandhoff disease, infantile, juvenile, and adult forms, 268800 |
| HFE | 100.0% | 100.0% | 100.0% | 98.3% | Hemochromatosis, type 1, 235200 |
| HGD | 100.0% | 99.7% | 100.0% | 98.6% | Alkaptonuria, 203500 |
| HGSNAT | 92.4% | 92.4% | 100.0% | 98.7% | Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930;Retinitis pigmentosa 73, 616544 |
| HIBADH | 100.0% | 100.0% | 100.0% | 97.9% | |
| HIBCH | 100.0% | 100.0% | 100.0% | 98.2% | 3-hydroxyisobutryl-CoA hydrolase deficiency, 250620 |

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| HK1 | 100.0% | 100.0% | 100.0% | 99.3% | Retinitis pigmentosa 79, 617460;Neuropathy, hereditary motor and sensory, Russe type, 605285;Neurodevelopmental disorder with visual defects and brain anomalies, 618547;Hemolytic anemia due to hexokinase deficiency, 235700 |
| HLCS | 100.0% | 100.0% | 99.9% | 97.6% | Holocarboxylase synthetase deficiency, 253270 |
| HMBS | 100.0% | 100.0% | 100.0% | 99.1% | Leukoencephalopathy, porphyria-related, 620711;Encephalopathy, porphyria-related, 620704;Porphyria, acute intermittent, nonerythroid variant, 176000;Porphyria, acute intermittent, 176000 |
| HMGCL | 100.0% | 100.0% | 100.0% | 98.6% | HMG-CoA lyase deficiency, 246450 |
| HMGCR | 100.0% | 100.0% | 100.0% | 99.0% | Muscular dystrophy, limb-girdle, autosomal recessive 28, 620375;[Statins, response to], 620410;[Low density lipoprotein cholesterol level QTL 3], 620410 |
| HMGCS2 | 100.0% | 100.0% | 100.0% | 99.0% | HMG-CoA synthase-2 deficiency, 605911 |

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| HMOX1 | 100.0% | 100.0% | 100.0% | 99.8% | Heme oxygenase-1 deficiency, 614034;{Pulmonary disease, chronic obstructive, susceptibility to}, 606963 |
| HNF1A | 100.0% | 100.0% | 100.0% | 99.6% | 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 |
| HNF4A | 100.0% | 100.0% | 100.0% | 99.3% | Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young, 616026;{Diabetes mellitus, noninsulin-dependent}, 125853;MODY, type I, 125850 |
| HOGA1 | 100.0% | 100.0% | 100.0% | 99.2% | Hyperoxaluria, primary, type III, 613616 |
| HPD | 100.0% | 100.0% | 100.0% | 97.0% | Hawkinsinuria, 140350;Tyrosinemia, type III, 276710 |

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| HPDL | 100.0% | 100.0% | 100.0% | 98.9% | Neurodevelopmental disorder with progressive spasticity and brain white matter abnormalities, 619026;Spastic paraplegia 83, autosomal recessive, 619027 |
| HPRT1 | 100.0% | 100.0% | 98.4% | 70.9% | Hyperuricemia, HRPT-related, 300323;Lesch-Nyhan syndrome, 300322 |
| HS6ST1 | 100.0% | 100.0% | 100.0% | 92.1% | {Hypogonadotropic hypogonadism 15 with or without anosmia}, 614880 |
| HSD11B1 | 100.0% | 100.0% | 100.0% | 99.6% | Cortisone reductase deficiency 2, 614662 |
| HSD11B2 | 100.0% | 100.0% | 99.9% | 94.8% | Apparent mineralocorticoid excess, 218030 |
| HSD17B10 | 100.0% | 99.8% | 98.0% | 70.3% | HSD10 mitochondrial disease, 300438 |
| HSD17B3 | 100.0% | 100.0% | 100.0% | 98.6% | Pseudohermaphroditism, male, with gynecomastia, 264300 |
| HSD17B4 | 100.0% | 100.0% | 100.0% | 98.2% | D-bifunctional protein deficiency, 261515;Perrault syndrome 1, 233400 |
| HSD3B2 | 99.6% | 99.4% | 100.0% | 98.8% | Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency, 201810 |

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| HSD3B7 | 100.0% | 100.0% | 100.0% | 99.9% | Bile acid synthesis defect, congenital, 1, 607765 |
| HTRA2 | 100.0% | 100.0% | 100.0% | 98.4% | {Parkinson disease 13}, 610297;3-methylglutaconic aciduria, type VIII, 617248 |
| HYAL1 | 100.0% | 100.0% | 100.0% | 98.2% | Mucopolysaccharidosis type IX, 601492 |
| IDH2 | 100.0% | 100.0% | 100.0% | 98.1% | D-2-hydroxyglutaric aciduria 2, 613657 |
| IDH3B | 100.0% | 100.0% | 100.0% | 99.0% | Retinitis pigmentosa 46, 612572 |
| IDI1 | 100.0% | 100.0% | 100.0% | 96.2% | |
| IDS | 100.0% | 100.0% | 97.1% | 69.8% | Mucopolysaccharidosis II, 309900 |
| IDUA | 100.0% | 100.0% | 100.0% | 97.9% | Mucopolysaccharidosis Is, 607016;Mucopolysaccharidosis Ih/s, 607015;Mucopolysaccharidosis Ih, 607014 |
| IMPDH1 | 100.0% | 100.0% | 100.0% | 98.5% | Retinitis pigmentosa 10, 180105;Leber congenital amaurosis 11, 613837 |
| INPP5E | 100.0% | 100.0% | 100.0% | 97.0% | Impaired intellectual development, truncal obesity, retinal dystrophy, and micropenis syndrome, 610156;Joubert syndrome 1, 213300 |
| INPPL1 | 100.0% | 100.0% | 100.0% | 98.8% | Opsismodysplasia, 258480 |

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| INSR | 100.0% | 100.0% | 100.0% | 98.4% | Rabson-Mendenhall syndrome, 262190;Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549;Donohue syndrome, 246200;Hyperinsulinemic hypoglycemia, familial, 5, 609968 |
| IREB2 | 100.0% | 100.0% | 100.0% | 98.8% | Neurodegeneration, early-onset, with choreoathetoid movements and microcytic anemia, 618451 |
| ITCH | 92.5% | 92.5% | 100.0% | 98.3% | Autoimmune disease, multisystem, with facial dysmorphism, 613385 |
| ITPA | 100.0% | 100.0% | 100.0% | 97.8% | [Inosine triphosphatase deficiency], 613850;Developmental and epileptic encephalopathy 35, 616647 |
| IVD | 100.0% | 100.0% | 100.0% | 99.2% | Isovaleric acidemia, 243500 |
| KCNA2 | 100.0% | 100.0% | 100.0% | 99.0% | Developmental and epileptic encephalopathy 32, 616366 |

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| KCNJ11 | 100.0% | 100.0% | 100.0% | 99.7% | Diabetes, permanent neonatal 2, with or without neurologic features, 618856;{Diabetes mellitus, type 2, susceptibility to}, 125853;Maturity-onset diabetes of the young, type 13, 616329;Diabetes mellitus, transient neonatal 3, 610582;Hyperinsulinemic hypoglycemia, familial, 2, 601820 |
| KMT2A | 99.2% | 99.2% | 100.0% | 97.9% | Wiedemann-Steiner syndrome, 605130 |
| KMT2D | 100.0% | 100.0% | 100.0% | 98.8% | Branchial arch abnormalities, choanal atresia, athelia, hearing loss, and hypothyroidism syndrome, 620186;Kabuki syndrome 1, 147920 |
| L2HGDH | 100.0% | 100.0% | 100.0% | 98.3% | L-2-hydroxyglutaric aciduria, 236792 |
| LAMP2 | 85.3% | 85.3% | 98.1% | 72.3% | Danon disease, 300257 |
| LARGE1 | 100.0% | 100.0% | 100.0% | 99.5% | 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 |

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| LCAT | 100.0% | 100.0% | 100.0% | 98.9% | Fish-eye disease, 136120;Norum disease, 245900 |
| LCT | 100.0% | 100.0% | 100.0% | 98.9% | Lactase deficiency, congenital, 223000 |
| LDHA | 100.0% | 100.0% | 100.0% | 98.4% | Glycogen storage disease XI, 612933 |
| LDHB | 100.0% | 100.0% | 100.0% | 98.4% | [Lactate dehydrogenase-B deficiency], 614128 |
| LFNG | 99.0% | 96.4% | 99.7% | 90.2% | Spondylocostal dysostosis 3, autosomal recessive, 609813 |
| LIAS | 100.0% | 100.0% | 100.0% | 99.2% | Hyperglycinemia, lactic acidosis, and seizures, 614462 |
| LIPA | 96.6% | 95.2% | 100.0% | 98.8% | Wolman disease, 620151;Cholesteryl ester storage disease, 278000 |
| LIPC | 100.0% | 100.0% | 100.0% | 99.5% | {Diabetes mellitus, noninsulin-dependent}, 125853;Hepatic lipase deficiency, 614025;[High density lipoprotein cholesterol level QTL 12], 612797 |
| LIPE | 100.0% | 100.0% | 100.0% | 99.0% | Lipodystrophy, familial partial, type 6, 615980 |
| LIPT1 | 100.0% | 100.0% | 100.0% | 96.6% | Lipoyltransferase 1 deficiency, 616299 |

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| LIPT2 | 100.0% | 100.0% | 100.0% | 98.2% | Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities, 617668 |
| LMBRD1 | 100.0% | 99.8% | 100.0% | 96.9% | Methylmalonic aciduria and homocystinuria, cblF type, 277380 |
| LMF1 | 100.0% | 100.0% | 100.0% | 99.1% | Lipase deficiency, combined, 246650 |
| LMNA | 100.0% | 100.0% | 100.0% | 99.2% | 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 |

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| LMNB2 | 100.0% | 99.8% | 100.0% | 97.3% | Microcephaly 27, primary, autosomal dominant, 619180;?Epilepsy, progressive myoclonic, 9, 616540;{Lipodystrophy, partial, acquired, susceptibility to}, 608709 |
| LPIN1 | 100.0% | 100.0% | 100.0% | 98.7% | Myoglobinuria, acute recurrent, autosomal recessive, 268200 |
| LPIN2 | 99.3% | 99.2% | 100.0% | 98.6% | Majeed syndrome, 609628 |
| LPL | 100.0% | 100.0% | 100.0% | 98.8% | Lipoprotein lipase deficiency, 238600;[High density lipoprotein cholesterol level QTL 11], 238600;Combined hyperlipidemia, familial, 144250 |
| LRAT | 100.0% | 100.0% | 100.0% | 98.5% | Leber congenital amaurosis 14, 613341;Retinal dystrophy, early-onset severe, 613341;Retinitis pigmentosa, juvenile, 613341 |
| LTC4S | 100.0% | 100.0% | 100.0% | 96.6% | Leukotriene C4 synthase deficiency, 614037 |
| LYST | 99.5% | 99.3% | 100.0% | 98.8% | Chediak-Higashi syndrome, 214500 |
| MAN1B1 | 100.0% | 99.1% | 100.0% | 99.4% | Rafiq syndrome, 614202 |
| MAN2B1 | 100.0% | 100.0% | 100.0% | 99.1% | Mannosidosis, alpha-, types I and II, 248500 |

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| MAN2B2 | 100.0% | 100.0% | 100.0% | 99.7% | |
| MAN2C1 | 100.0% | 100.0% | 100.0% | 99.3% | Congenital disorder of deglycosylation 2, 619775 |
| MANBA | 100.0% | 100.0% | 100.0% | 98.2% | Mannosidosis, beta, 248510 |
| MAOA | 99.4% | 98.5% | 98.1% | 72.6% | {Antisocial behavior}, 300615;Brunner syndrome, 300615 |
| MAT1A | 100.0% | 100.0% | 100.0% | 99.5% | Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850;Methionine adenosyltransferase deficiency, autosomal recessive, 250850 |
| MBOAT7 | 100.0% | 100.0% | 100.0% | 98.3% | Intellectual developmental disorder, autosomal recessive 57, 617188 |
| MCCC1 | 100.0% | 100.0% | 100.0% | 99.1% | 3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200 |
| MCCC2 | 93.4% | 93.4% | 100.0% | 97.6% | 3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210 |
| MCEE | 100.0% | 100.0% | 100.0% | 98.5% | Methylmalonyl-CoA epimerase deficiency, 251120 |

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| MCOLN1 | 100.0% | 100.0% | 100.0% | 99.3% | Lisch epithelial corneal dystrophy, 620763;Mucolipidosis IV, 252650 |
| MDH1 | 100.0% | 100.0% | 100.0% | 99.2% | ?Developmental and epileptic encephalopathy 88, 618959 |
| MFSD2A | 100.0% | 100.0% | 100.0% | 98.9% | Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain abnormalities, 616486 |
| MFSD8 | 100.0% | 100.0% | 100.0% | 99.2% | Macular dystrophy with central cone involvement, 616170;Ceroid lipofuscinosis, neuronal, 7, 610951 |
| MGAT2 | 100.0% | 100.0% | 100.0% | 97.5% | Congenital disorder of glycosylation, type IIa, 212066 |
| MINPP1 | 100.0% | 100.0% | 100.0% | 98.1% | {Thyroid carcinoma, follicular}, 188470;Pontocerebellar hypoplasia, type 16, 619527 |
| MLYCD | 100.0% | 100.0% | 100.0% | 97.4% | Malonyl-CoA decarboxylase deficiency, 248360 |
| MMAA | 100.0% | 100.0% | 100.0% | 99.1% | Methylmalonic aciduria, vitamin B12-responsive, cblA type, 251100 |
| MMAB | 100.0% | 100.0% | 99.9% | 97.9% | Methylmalonic aciduria, vitamin B12-responsive, cblB type, 251110 |

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| MMACHC | 100.0% | 100.0% | 100.0% | 98.8% | Methylmalonic aciduria and homocystinuria, cblC type, 277400 |
| MMADHC | 89.3% | 89.3% | 100.0% | 98.1% | Methylmalonic aciduria, cblD type, variant 2, 277410;Methylmalonic aciduria and homocystinuria, cblD type, 277410;Homocystinuria, cblD type, variant 1, 277410 |
| MMS19 | 100.0% | 100.0% | 100.0% | 99.4% | |
| MMUT | 100.0% | 100.0% | 100.0% | 98.5% | Methylmalonic aciduria, mut(0) type, 251000 |
| MOCOS | 100.0% | 100.0% | 100.0% | 98.7% | Xanthinuria, type II, 603592 |
| MOCS1 | 100.0% | 100.0% | 100.0% | 98.8% | Molybdenum cofactor deficiency A, 252150 |
| MOCS2 | 100.0% | 100.0% | 100.0% | 98.5% | Molybdenum cofactor deficiency B, 252160 |
| MOGS | 100.0% | 100.0% | 100.0% | 99.6% | Congenital disorder of glycosylation, type IIb, 606056 |
| MORC2 | 100.0% | 100.0% | 100.0% | 98.5% | Charcot-Marie-Tooth disease, axonal, type 2Z, 616688;Developmental delay, impaired growth, dysmorphic facies, and axonal neuropathy, 619090 |
| MPC2 | 100.0% | 100.0% | 100.0% | 96.0% | |
| MPDU1 | 100.0% | 100.0% | 100.0% | 97.2% | Congenital disorder of glycosylation, type If, 609180 |

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| MPI | 100.0% | 100.0% | 100.0% | 99.7% | Congenital disorder of glycosylation, type Ib, 602579 |
| MRPL44 | 100.0% | 100.0% | 100.0% | 98.9% | Combined oxidative phosphorylation deficiency 16, 615395 |
| MRPS36 | 100.0% | 100.0% | 100.0% | 96.9% | |
| MSMO1 | 100.0% | 100.0% | 100.0% | 99.3% | Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834 |
| MTHFD1 | 100.0% | 100.0% | 100.0% | 99.1% | {Neural tube defects, folate-sensitive, susceptibility to}, 601634;Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia, 617780 |
| MTHFR | 100.0% | 100.0% | 100.0% | 98.3% | Homocystinuria due to MTHFR deficiency, 236250;{Thromboembolism, susceptibility to}, 188050;{Schizophrenia, susceptibility to}, 181500;{Neural tube defects, susceptibility to}, 601634;{Vascular disease, susceptibility to}, |
| MTM1 | 99.7% | 99.2% | 97.6% | 70.5% | Myopathy, centronuclear, X-linked, 310400 |
| MTMR2 | 100.0% | 100.0% | 99.9% | 98.6% | Charcot-Marie-Tooth disease, type 4B1, 601382 |

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| MTR | 100.0% | 100.0% | 100.0% | 98.2% | {Neural tube defects, folate-sensitive, susceptibility to}, 601634;Homocystinuria-megaloblastic anemia, cblG complementation type, 250940 |
| MTRR | 100.0% | 100.0% | 100.0% | 98.4% | Homocystinuria-megaloblastic anemia, cbl E type, 236270;{Neural tube defects, folate-sensitive, susceptibility to}, 601634 |
| MVK | 100.0% | 100.0% | 100.0% | 99.7% | Hyper-IgD syndrome, 260920;Porokeratosis 3, multiple types, 175900;Mevalonic aciduria, 610377 |
| NADK2 | 100.0% | 100.0% | 100.0% | 95.8% | 2,4-dienoyl-CoA reductase deficiency, 616034 |
| NAGA | 100.0% | 100.0% | 100.0% | 99.5% | Schindler disease, type I, 609241;Kanzaki disease, 609242;Schindler disease, type III, 609241 |
| NAGLU | 100.0% | 100.0% | 100.0% | 98.4% | ?Charcot-Marie-Tooth disease, axonal, type 2V, 616491;Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920 |
| NAGS | 100.0% | 100.0% | 100.0% | 98.4% | N-acetylglutamate synthase deficiency, 237310 |
| NANS | 100.0% | 100.0% | 100.0% | 98.1% | Spondyloepimetaphyseal dysplasia, Genevieve type, 610442 |

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| NAXD | 96.8% | 92.4% | 100.0% | 99.2% | Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 2, 618321 |
| NAXE | 95.8% | 91.2% | 100.0% | 98.5% | Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186 |
| NBAS | 100.0% | 99.8% | 100.0% | 98.7% | Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800;Infantile liver failure syndrome 2, 616483 |
| NEU1 | 100.0% | 100.0% | 100.0% | 99.4% | Sialidosis, type II, 256550;Sialidosis, type I, 256550 |
| NGLY1 | 100.0% | 100.0% | 100.0% | 98.7% | Congenital disorder of deglycosylation 1, 615273 |
| NMNAT1 | 99.9% | 97.7% | 100.0% | 97.0% | Spondyloepiphyseal dysplasia, sensorineural hearing loss, intellectual developmental disorder, and Leber congenital amaurosis, 619260;Leber congenital amaurosis 9, 608553 |
| NNT | 96.4% | 96.3% | 100.0% | 99.2% | Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency, 614736 |

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| NPC1 | 100.0% | 100.0% | 100.0% | 99.1% | Niemann-Pick disease, type C1, 257220;Niemann-Pick disease, type D, 257220 |
| NPC2 | 100.0% | 100.0% | 100.0% | 98.5% | Niemann-pick disease, type C2, 607625 |
| NPL | 100.0% | 100.0% | 100.0% | 98.9% | |
| NSD1 | 100.0% | 100.0% | 100.0% | 98.6% | Sotos syndrome, 117550 |
| NSDHL | 100.0% | 99.9% | 99.5% | 74.7% | CK syndrome, 300831;CHILD syndrome, 308050 |
| NT5C3A | 100.0% | 100.0% | 100.0% | 98.1% | Anemia, hemolytic, due to UMPH1 deficiency, 266120 |
| NT5E | 100.0% | 100.0% | 100.0% | 98.1% | Calcification of joints and arteries, 211800 |
| NUS1 | 100.0% | 100.0% | 100.0% | 98.7% | Intellectual developmental disorder, autosomal dominant 55, with seizures, 617831;?Congenital disorder of glycosylation, type 1aa, 617082 |
| OAT | 100.0% | 100.0% | 100.0% | 98.2% | Gyrate atrophy of choroid and retina with or without ornithinemia, 258870 |
| OCRL | 100.0% | 100.0% | 97.8% | 69.7% | Dent disease 2, 300555;Lowe syndrome, 309000 |
| ODC1 | 100.0% | 100.0% | 100.0% | 99.0% | Bachmann-Bupp syndrome, 619075 |

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| OGDH | 100.0% | 100.0% | 100.0% | 99.3% | Oxoglutarate dehydrogenase deficiency, 203740 |
| OGDHL | 100.0% | 100.0% | 100.0% | 99.2% | Yoon-Bellen neurodevelopmental syndrome, 619701 |
| OPA3 | 100.0% | 100.0% | 100.0% | 98.6% | 3-methylglutaconic aciduria, type III, 258501;Optic atrophy 3 with cataract, 165300 |
| OPLAH | 100.0% | 100.0% | 100.0% | 98.9% | 5-oxoprolinase deficiency, 260005 |
| OTC | 100.0% | 99.6% | 96.8% | 67.8% | Ornithine transcarbamylase deficiency, 311250 |
| OXCT1 | 100.0% | 100.0% | 100.0% | 97.8% | Succinyl CoA:3-oxoacid CoA transferase deficiency, 245050 |
| PAH | 100.0% | 100.0% | 100.0% | 99.2% | [Hyperphenylalaninemia, non-PKU mild], 261600;Phenylketonuria, 261600 |
| PANK2 | 100.0% | 100.0% | 100.0% | 98.6% | Neurodegeneration with brain iron accumulation 1, 234200 |
| PC | 100.0% | 100.0% | 100.0% | 99.7% | Pyruvate carboxylase deficiency, 266150 |
| PCBD1 | 100.0% | 100.0% | 100.0% | 99.4% | Hyperphenylalaninemia, BH4-deficient, D, 264070 |
| PCCA | 100.0% | 100.0% | 100.0% | 98.4% | Propionicacidemia, 606054 |
| PCCB | 99.9% | 98.0% | 100.0% | 97.9% | Propionicacidemia, 606054 |

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|---|---|---|---|---|---|
| PCK1 | 100.0% | 100.0% | 100.0% | 99.2% | Phosphoenolpyruvate carboxykinase deficiency, cytosolic, 261680 |
| PCK2 | 100.0% | 100.0% | 100.0% | 99.2% | PEPCK deficiency, mitochondrial, 261650 |
| PCYT1A | 100.0% | 100.0% | 100.0% | 98.6% | Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940;Lipodystrophy, congenital generalized, type 5, 620680 |
| PCYT2 | 100.0% | 100.0% | 99.9% | 97.8% | Spastic paraplegia 82, autosomal recessive, 618770 |
| PDSS1 | 100.0% | 100.0% | 100.0% | 97.3% | Coenzyme Q10 deficiency, primary, 2, 614651 |
| PDSS2 | 100.0% | 100.0% | 100.0% | 98.5% | Coenzyme Q10 deficiency, primary, 3, 614652 |
| PEPD | 93.9% | 93.9% | 100.0% | 99.5% | Prolidase deficiency, 170100 |
| PEX1 | 100.0% | 100.0% | 100.0% | 98.5% | Heimler syndrome 1, 234580;Peroxisome biogenesis disorder 1B (NALD/IRD), 601539;Peroxisome biogenesis disorder 1A (Zellweger), 214100 |
| PEX10 | 100.0% | 100.0% | 100.0% | 99.8% | Peroxisome biogenesis disorder 6A (Zellweger), 614870;Peroxisome biogenesis disorder 6B, 614871 |

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| PEX11B | 100.0% | 100.0% | 100.0% | 96.3% | Peroxisome biogenesis disorder 14B, 614920 |
| PEX12 | 100.0% | 100.0% | 100.0% | 98.8% | Peroxisome biogenesis disorder 3B, 266510;Peroxisome biogenesis disorder 3A (Zellweger), 614859 |
| PEX13 | 100.0% | 100.0% | 100.0% | 97.8% | Peroxisome biogenesis disorder 11A (Zellweger), 614883;Peroxisome biogenesis disorder 11B, 614885 |
| PEX14 | 100.0% | 100.0% | 100.0% | 99.1% | Peroxisome biogenesis disorder 13A (Zellweger), 614887 |
| PEX16 | 100.0% | 100.0% | 100.0% | 99.2% | Peroxisome biogenesis disorder 8B, 614877;Peroxisome biogenesis disorder 8A (Zellweger), 614876 |
| PEX19 | 100.0% | 100.0% | 100.0% | 99.0% | Peroxisome biogenesis disorder 12A (Zellweger), 614886 |
| PEX2 | 100.0% | 100.0% | 100.0% | 98.9% | Peroxisome biogenesis disorder 5A (Zellweger), 614866;Peroxisome biogenesis disorder 5B, 614867 |

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| PEX26 | 100.0% | 100.0% | 100.0% | 98.0% | Peroxisome biogenesis disorder 7B, 614873;Peroxisome biogenesis disorder 7A (Zellweger), 614872 |
| PEX3 | 100.0% | 100.0% | 100.0% | 97.9% | Peroxisome biogenesis disorder 10A (Zellweger), 614882;?Peroxisome biogenesis disorder 10B, 617370 |
| PEX5 | 100.0% | 100.0% | 100.0% | 98.8% | Peroxisome biogenesis disorder 2B, 202370;Peroxisome biogenesis disorder 2A (Zellweger), 214110;Rhizomelic chondrodysplasia punctata, type 5, 616716 |
| PEX6 | 100.0% | 100.0% | 100.0% | 97.9% | Peroxisome biogenesis disorder 4B, 614863;Peroxisome biogenesis disorder 4A (Zellweger), 614862;Heimler syndrome 2, 616617 |
| PEX7 | 97.9% | 97.9% | 100.0% | 98.8% | Rhizomelic chondrodysplasia punctata, type 1, 215100;Peroxisome biogenesis disorder 9B, 614879 |
| PFKM | 100.0% | 100.0% | 100.0% | 99.2% | Glycogen storage disease VII, 232800 |

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| PGAM2 | 100.0% | 100.0% | 100.0% | 99.0% | Glycogen storage disease X, 261670 |
| PGAP1 | 100.0% | 100.0% | 100.0% | 97.9% | Neurodevelopmental disorder with dysmorphic features, spasticity, and brain abnormalities, 615802 |
| PGAP2 | 100.0% | 100.0% | 100.0% | 98.7% | Hyperphosphatasia with impaired intellectual development syndrome 3, 614207 |
| PGAP3 | 100.0% | 100.0% | 100.0% | 99.4% | Hyperphosphatasia with impaired intellectual development syndrome 4, 615716 |
| PGK1 | 100.0% | 99.7% | 98.3% | 72.9% | Phosphoglycerate kinase 1 deficiency, 300653 |
| PGM1 | 94.0% | 94.0% | 100.0% | 98.0% | Congenital disorder of glycosylation, type It, 614921 |
| PGM2L1 | 100.0% | 100.0% | 100.0% | 98.6% | Neurodevelopmental disorder with hypotonia, dysmorphic facies, and skin abnormalities, 620191 |
| PGM3 | 100.0% | 100.0% | 100.0% | 99.2% | Immunodeficiency 23, 615816 |
| PHGDH | 100.0% | 100.0% | 100.0% | 99.2% | Neu-Laxova syndrome 1, 256520;Phosphoglycerate dehydrogenase deficiency, 601815 |
| PHKA1 | 100.0% | 100.0% | 97.6% | 71.6% | Muscle glycogenosis, 300559 |

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| PHKA2 | 100.0% | 100.0% | 98.3% | 72.6% | Glycogen storage disease, type IXa2, 306000;Glycogen storage disease, type IXa1, 306000 |
| PHKB | 100.0% | 100.0% | 100.0% | 98.7% | Phosphorylase kinase deficiency of liver and muscle, autosomal recessive, 261750 |
| PHKG1 | 100.0% | 100.0% | 100.0% | 99.3% | |
| PHKG2 | 100.0% | 100.0% | 99.9% | 98.7% | Glycogen storage disease IXc, 613027 |
| PHYH | 100.0% | 100.0% | 100.0% | 98.2% | Refsum disease, 266500 |
| PI4K2A | 100.0% | 100.0% | 100.0% | 97.1% | Neurodevelopmental disorder with hyperkinetic movements, seizures and structural brain abnormalities, 620732 |
| PI4KA | 100.0% | 99.8% | 99.9% | 98.2% | Spastic paraplegia 84, autosomal recessive, 619621;Gastrointestinal defects and immunodeficiency syndrome 2, 619708;Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis, 616531 |

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| PIGA | 100.0% | 100.0% | 97.7% | 73.6% | Paroxysmal nocturnal hemoglobinuria, somatic, 300818;Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868;Neurodevelopmental disorder with epilepsy and hemochromatosis, 301072 |
| PIGB | 100.0% | 100.0% | 100.0% | 97.7% | Developmental and epileptic encephalopathy 80, 618580 |
| PIGC | 100.0% | 100.0% | 100.0% | 99.7% | Glycosylphosphatidylinositol biosynthesis defect 16, 617816 |
| PIGL | 100.0% | 100.0% | 100.0% | 98.2% | CHIME syndrome, 280000 |
| PIGM | 100.0% | 100.0% | 100.0% | 98.4% | Glycosylphosphatidylinositol deficiency, 610293 |
| PIGN | 100.0% | 99.9% | 100.0% | 98.6% | Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080 |
| PIGO | 100.0% | 100.0% | 100.0% | 99.2% | Hyperphosphatasia with impaired intellectual development syndrome 2, 614749 |
| PIGP | 100.0% | 100.0% | 100.0% | 96.8% | Developmental and epileptic encephalopathy 55, 617599 |
| PIGQ | 100.0% | 100.0% | 100.0% | 99.2% | Multiple congenital anomalies-hypotonia-seizures syndrome 4, 618548 |

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| PIGT | 100.0% | 100.0% | 100.0% | 99.3% | ?Paroxysmal nocturnal hemoglobinuria 2, 615399;Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398 |
| PIGV | 100.0% | 99.6% | 100.0% | 99.3% | Hyperphosphatasia with impaired intellectual development syndrome 1, 239300 |
| PIGW | 100.0% | 100.0% | 100.0% | 98.9% | Glycosylphosphatidylinositol biosynthesis defect 11, 616025 |
| PIGY | 100.0% | 100.0% | 100.0% | 99.1% | Hyperphosphatasia with impaired intellectual development syndrome 6, 616809 |

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| PIK3CA | 100.0% | 100.0% | 100.0% | 98.0% | 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;Nevus, epidermal, somatic, 162900;Gastric cancer, somatic, 613659;Nonsmall cell lung cancer, somatic, 211980;Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501;Cowden syndrome 5, 615108 |
| PIK3R1 | 100.0% | 99.8% | 100.0% | 98.4% | Immunodeficiency 36, 616005;?Agammaglobulinemia 7, autosomal recessive, 615214;SHORT syndrome, 269880 |

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| PIK3R2 | 100.0% | 100.0% | 100.0% | 97.5% | Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387 |
| PIK3R5 | 100.0% | 100.0% | 100.0% | 99.4% | Ataxia-oculomotor apraxia 3, 615217 |
| PIKFYVE | 100.0% | 100.0% | 100.0% | 98.5% | Corneal fleck dystrophy, 121850 |
| PIP5K1C | 100.0% | 100.0% | 100.0% | 98.8% | Lethal congenital contractural syndrome 3, 611369 |
| PKLR | 100.0% | 100.0% | 100.0% | 99.4% | Adenosine triphosphate, elevated, of erythrocytes, 102900;Pyruvate kinase deficiency, 266200 |
| PLA2G5 | 100.0% | 100.0% | 100.0% | 98.7% | [Fleck retina, familial benign], 228980 |
| PLA2G6 | 100.0% | 99.9% | 100.0% | 99.2% | Parkinson disease 14, autosomal recessive, 612953;Neurodegeneration with brain iron accumulation 2B, 610217;Infantile neuroaxonal dystrophy 1, 256600 |
| PLA2G7 | 100.0% | 100.0% | 100.0% | 96.7% | Platelet-activating factor acetylhydrolase deficiency, 614278 |
| PLAAT3 | 100.0% | 100.0% | 100.0% | 99.6% | Lipodystrophy, familial partial, type 9, 620683 |
| PLCB1 | 100.0% | 100.0% | 100.0% | 97.7% | Developmental and epileptic encephalopathy 12, 613722 |

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| PLCB4 | 99.0% | 98.9% | 100.0% | 98.3% | Auriculocondylar syndrome 2B, 620458;Auriculocondylar syndrome 2A, 614669 |
| PLCD1 | 100.0% | 100.0% | 100.0% | 99.6% | Nail disorder, nonsyndromic congenital, 3, (leukonychia), 151600 |
| PLCE1 | 100.0% | 99.8% | 100.0% | 98.5% | Nephrotic syndrome, type 3, 610725 |
| PLCG2 | 100.0% | 100.0% | 100.0% | 99.0% | Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878;Familial cold autoinflammatory syndrome 3, 614468 |
| PLIN1 | 100.0% | 100.0% | 100.0% | 98.4% | Lipodystrophy, familial partial, type 4, 613877 |
| PLOD1 | 100.0% | 100.0% | 100.0% | 98.5% | Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400 |
| PLOD2 | 100.0% | 100.0% | 99.9% | 97.0% | Bruck syndrome 2, 609220 |
| PLOD3 | 100.0% | 100.0% | 100.0% | 98.0% | BCARD syndrome (lysyl hydroxylase 3 deficiency), 612394 |
| PLPBP | 100.0% | 100.0% | 100.0% | 99.2% | Epilepsy, early-onset, 1, vitamin B6-dependent, 617290 |
| PMM2 | 100.0% | 100.0% | 100.0% | 98.2% | Congenital disorder of glycosylation, type Ia, 212065 |

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| PNLIP | 100.0% | 100.0% | 100.0% | 98.7% | ?Pancreatic lipase deficiency, 614338 |
| PNMT | 100.0% | 100.0% | 100.0% | 97.2% | |
| PNP | 100.0% | 100.0% | 100.0% | 99.0% | Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179 |
| PNPLA2 | 100.0% | 100.0% | 100.0% | 99.5% | Neutral lipid storage disease with myopathy, 610717 |
| PNPLA6 | 100.0% | 100.0% | 100.0% | 99.7% | Spastic paraplegia 39, autosomal recessive, 612020;Oliver-McFarlane syndrome, 275400;?Laurence-Moon syndrome, 245800;Boucher-Neuhauser syndrome, 215470 |
| PNPO | 100.0% | 100.0% | 100.0% | 99.1% | Pyridoxamine 5'-phosphate oxidase deficiency, 610090 |
| POFUT1 | 100.0% | 100.0% | 100.0% | 98.7% | Dowling-Degos disease 2, 615327 |
| POGLUT1 | 100.0% | 100.0% | 100.0% | 98.8% | Dowling-Degos disease 4, 615696;Muscular dystrophy, limb-girdle, autosomal recessive 21, 617232 |

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| POLD1 | 100.0% | 100.0% | 100.0% | 99.2% | Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381;Immunodeficiency 120, 620836;{Colorectal cancer, susceptibility to, 10}, 612591 |
| POLR3A | 100.0% | 100.0% | 100.0% | 98.8% | Wiedemann-Rautenstrauch syndrome, 264090;Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 |
| POLR3B | 100.0% | 99.9% | 100.0% | 98.3% | 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 | 100.0% | 100.0% | 100.0% | 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 | 100.0% | 100.0% | 100.0% | 99.9% | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 8, 614830;Muscular dystrophy-dystroglycanopathy (limb-girdle) type C, 8, 618135 |
| POMK | 100.0% | 100.0% | 100.0% | 99.8% | ?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249 |

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| POMT1 | 100.0% | 100.0% | 100.0% | 98.1% | 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 |
| POMT2 | 100.0% | 100.0% | 100.0% | 96.5% | 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 |

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| PPARG | 99.9% | 99.6% | 100.0% | 99.1% | {Diabetes, type 2}, 125853;Insulin resistance, severe, digenic, 604367;Lipodystrophy, familial partial, type 3, 604367;Obesity, severe, 601665;Carotid intimal medial thickness 1, 609338;[Obesity, resistance to], |
| PPCDC | 100.0% | 100.0% | 100.0% | 99.0% | |
| PPCS | 100.0% | 100.0% | 100.0% | 98.6% | Cardiomyopathy, dilated, 2C, 618189 |
| PPFIA3 | 100.0% | 100.0% | 100.0% | 98.1% | |
| PPM1K | 100.0% | 100.0% | 100.0% | 99.4% | Maple syrup urine disease, mild variant, 615135 |
| PPOX | 100.0% | 100.0% | 100.0% | 98.9% | Variegate porphyria, childhood-onset, 620483;Variegate porphyria, 176200 |
| PPT1 | 90.3% | 90.3% | 100.0% | 97.8% | Ceroid lipofuscinosis, neuronal, 1, 256730 |
| PRKAG2 | 100.0% | 100.0% | 100.0% | 96.5% | Glycogen storage disease of heart, lethal congenital, 261740;Wolff-Parkinson-White syndrome, 194200;Cardiomyopathy, hypertrophic 6, 600858 |
| PRKCSH | 100.0% | 100.0% | 100.0% | 98.7% | Polycystic liver disease 1, 174050 |

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| PRODH | 100.0% | 100.0% | 100.0% | 99.3% | {Schizophrenia, susceptibility to, 4}, 600850;Hyperprolinemia, type I, 239500 |
| PRPS1 | 100.0% | 100.0% | 96.3% | 69.8% | Arts syndrome, 301835;Phosphoribosylpyrophosphate synthetase superactivity, 300661;Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070;Deafness, X-linked 1, 304500;Gout, PRPS-related, 300661 |
| PSAP | 100.0% | 100.0% | 100.0% | 99.1% | 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 | 100.0% | 100.0% | 100.0% | 98.3% | Neu-Laxova syndrome 2, 616038;Phosphoserine aminotransferase deficiency, 610992 |
| PSPH | 100.0% | 100.0% | 100.0% | 98.5% | Phosphoserine phosphatase deficiency, 614023 |

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| PTEN | 94.5% | 94.5% | 99.8% | 93.1% | {Glioma susceptibility 2}, 613028;{Meningioma}, 607174;Cowden syndrome 1, 158350;Lhermitte-Duclos disease, 158350;Prostate cancer, somatic, 176807;Macrocephaly/autism syndrome, 605309 |
| PTGIS | 100.0% | 100.0% | 100.0% | 98.9% | Hypertension, essential, 145500 |
| PTPN11 | 89.3% | 89.2% | 100.0% | 98.3% | Noonan syndrome 1, 163950;LEOPARD syndrome 1, 151100;Metachondromatosis, 156250;Leukemia, juvenile myelomonocytic, somatic, 607785 |
| PTS | 100.0% | 100.0% | 100.0% | 95.8% | Hyperphenylalaninemia, BH4-deficient, A, 261640 |
| PUS3 | 100.0% | 100.0% | 100.0% | 99.3% | Neurodevelopmental disorder with microcephaly and gray sclerae, 617051 |
| PYCR1 | 100.0% | 100.0% | 100.0% | 99.8% | Cutis laxa, autosomal recessive, type IIIB, 614438;Cutis laxa, autosomal recessive, type IIB, 612940 |
| PYCR2 | 100.0% | 100.0% | 100.0% | 98.6% | Leukodystrophy, hypomyelinating, 10, 616420 |
| PYGL | 100.0% | 100.0% | 100.0% | 99.2% | Glycogen storage disease VI, 232700 |

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| PYGM | 100.0% | 100.0% | 100.0% | 99.6% | McArdle disease, 232600 |
| QDPR | 100.0% | 100.0% | 100.0% | 97.6% | Hyperphenylalaninemia, BH4-deficient, C, 261630 |
| RBCK1 | 100.0% | 100.0% | 99.9% | 97.8% | Polyglucosan body myopathy 1 with or without immunodeficiency, 615895 |
| RDH12 | 100.0% | 100.0% | 100.0% | 99.4% | Leber congenital amaurosis 13, 612712 |
| RDH5 | 100.0% | 100.0% | 100.0% | 99.3% | Fundus albipunctatus, 136880 |
| RFT1 | 100.0% | 100.0% | 100.0% | 98.8% | Congenital disorder of glycosylation, type In, 612015 |
| RINT1 | 100.0% | 100.0% | 100.0% | 98.6% | Infantile liver failure syndrome 3, 618641 |
| RPE65 | 100.0% | 100.0% | 100.0% | 98.3% | Retinitis pigmentosa 20, 613794;Retinitis pigmentosa 87 with choroidal involvement, 618697;Leber congenital amaurosis 2, 204100 |
| RPIA | 100.0% | 100.0% | 100.0% | 98.2% | Ribose 5-phosphate isomerase deficiency, 608611 |
| RPN2 | 100.0% | 100.0% | 100.0% | 99.1% | |
| RXYLT1 | 100.0% | 100.0% | 100.0% | 96.8% | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041 |

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| SARDH | 91.7% | 91.7% | 100.0% | 98.5% | [Sarcosinemia], 268900 |
| SAT1 | 100.0% | 100.0% | 97.1% | 66.3% | |
| SC5D | 100.0% | 100.0% | 100.0% | 98.5% | Lathosterolosis, 607330 |
| SCARB2 | 100.0% | 100.0% | 100.0% | 99.1% | Epilepsy, progressive myoclonic 4, with or without renal failure, 254900 |
| SCP2 | 100.0% | 100.0% | 100.0% | 97.9% | ?Leukoencephalopathy with dystonia and motor neuropathy, 613724 |
| SCYL1 | 100.0% | 100.0% | 100.0% | 98.1% | Spinocerebellar ataxia, autosomal recessive 21, 616719 |
| SEC23B | 100.0% | 100.0% | 100.0% | 98.4% | ?Cowden syndrome 7, 616858;Dyserythropoietic anemia, congenital, type II, 224100 |
| SELENBP1 | 100.0% | 100.0% | 100.0% | 99.4% | Extraoral halitosis due to MTO deficiency, 618148 |
| SEPHS1 | 100.0% | 100.0% | 100.0% | 99.6% | |
| SEPSECS | 98.6% | 94.4% | 100.0% | 98.2% | Pontocerebellar hypoplasia type 2D, 613811 |
| SERAC1 | 100.0% | 100.0% | 100.0% | 98.3% | 3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739 |
| SGMS1 | 100.0% | 100.0% | 100.0% | 98.8% | |
| SGSH | 100.0% | 100.0% | 100.0% | 99.7% | Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900 |

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| SHMT2 | 100.0% | 100.0% | 100.0% | 99.6% | Neurodevelopmental disorder with cardiomyopathy, spasticity, and brain abnormalities, 619121 |
| SI | 99.0% | 98.3% | 100.0% | 98.5% | Sucrase-isomaltase deficiency, congenital, 222900 |
| SLC10A7 | 92.8% | 92.8% | 100.0% | 98.8% | Short stature, amelogenesis imperfecta, and skeletal dysplasia with scoliosis, 618363 |
| SLC12A1 | 96.4% | 96.3% | 100.0% | 98.4% | Bartter syndrome, type 1, 601678 |
| SLC13A3 | 100.0% | 100.0% | 100.0% | 99.3% | Leukoencephalopathy, acute reversible, with increased urinary alpha-ketoglutarate, 618384 |
| SLC16A1 | 100.0% | 100.0% | 100.0% | 99.7% | Hyperinsulinemic hypoglycemia, familial, 7, 610021;Erythrocyte lactate transporter defect, 245340;Monocarboxylate transporter 1 deficiency, 616095 |
| SLC17A5 | 100.0% | 100.0% | 100.0% | 97.2% | Salla disease, 604369;Sialic acid storage disorder, infantile, 269920 |
| SLC18A2 | 100.0% | 100.0% | 100.0% | 98.7% | Parkinsonism-dystonia, infantile, 2, 618049 |

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| SLC1A1 | 100.0% | 100.0% | 100.0% | 98.7% | Dicarboxylic aminoaciduria, 222730;{?Schizophrenia susceptibility 18}, 615232 |
| SLC1A4 | 100.0% | 100.0% | 100.0% | 98.2% | Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657 |
| SLC22A12 | 100.0% | 99.8% | 99.9% | 97.2% | Hypouricemia, renal, 220150 |
| SLC22A5 | 100.0% | 100.0% | 100.0% | 98.3% | Carnitine deficiency, systemic primary, 212140 |
| SLC25A1 | 100.0% | 100.0% | 100.0% | 93.2% | Combined D-2- and L-2-hydroxyglutaric aciduria, 615182;Myasthenic syndrome, congenital, 23, presynaptic, 618197 |
| SLC25A13 | 100.0% | 100.0% | 100.0% | 98.7% | Citrullinemia, type II, neonatal-onset, 605814;Citrullinemia, adult-onset type II, 603471 |
| SLC25A15 | 100.0% | 100.0% | 100.0% | 99.1% | Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970 |
| SLC25A19 | 100.0% | 100.0% | 100.0% | 98.7% | Microcephaly, Amish type, 607196;Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710 |

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| SLC25A20 | 100.0% | 100.0% | 100.0% | 99.2% | Carnitine-acylcarnitine translocase deficiency, 212138 |
| SLC25A21 | 100.0% | 100.0% | 100.0% | 98.5% | ?Mitochondrial DNA depletion syndrome 18, 618811 |
| SLC25A32 | 100.0% | 100.0% | 100.0% | 98.9% | ?Exercise intolerance, riboflavin-responsive, 616839 |
| SLC25A36 | 100.0% | 100.0% | 100.0% | 97.2% | Hyperinsulinemic hypoglycemia, familial, 8, 620211 |
| SLC25A38 | 100.0% | 100.0% | 100.0% | 99.2% | Anemia, sideroblastic, 2, pyridoxine-refractory, 205950 |
| SLC25A42 | 100.0% | 100.0% | 100.0% | 99.2% | Metabolic crises, recurrent, with variable encephalomyopathic features and neurologic regression, 618416 |
| SLC28A1 | 100.0% | 100.0% | 100.0% | 98.9% | [Uridine-cytidineuria], 618477 |

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| SLC2A1 | 100.0% | 100.0% | 100.0% | 99.4% | 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 | 100.0% | 100.0% | 100.0% | 99.4% | Fanconi-Bickel syndrome, 227810;{Diabetes mellitus, noninsulin-dependent}, 125853 |
| SLC2A9 | 100.0% | 100.0% | 100.0% | 98.9% | {Uric acid concentration, serum, QTL 2}, 612076;Hypouricemia, renal, 2, 612076 |
| SLC30A10 | 100.0% | 100.0% | 100.0% | 98.5% | Hypermanganesemia with dystonia 1, 613280 |
| SLC33A1 | 100.0% | 100.0% | 100.0% | 97.5% | Spastic paraplegia 42, autosomal dominant, 612539;Huppke-Brendel syndrome, 614482 |
| SLC35A1 | 100.0% | 100.0% | 100.0% | 99.2% | Congenital disorder of glycosylation, type IIf, 603585 |
| SLC35A2 | 100.0% | 100.0% | 98.8% | 74.8% | Congenital disorder of glycosylation, type IIm, 300896 |

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| SLC35A3 | 97.7% | 93.3% | 99.9% | 96.4% | Arthrogryposis, impaired intellectual development, and seizures, 615553 |
| SLC35C1 | 100.0% | 100.0% | 100.0% | 99.8% | Congenital disorder of glycosylation, type IIc, 266265 |
| SLC35D1 | 100.0% | 100.0% | 100.0% | 97.5% | Schneckenbecken dysplasia, 269250 |
| SLC36A2 | 100.0% | 100.0% | 100.0% | 98.7% | [Iminoglycinuria], 242600;[Hyperglycinuria], 138500 |
| SLC37A4 | 100.0% | 100.0% | 100.0% | 99.7% | Glycogen storage disease Ib, 232220;Congenital disorder of glycosylation, type IIw, 619525;Glycogen storage disease Ic, 232240 |
| SLC38A3 | 100.0% | 100.0% | 100.0% | 98.7% | Developmental and epileptic encephalopathy 102, 619881 |
| SLC39A14 | 93.6% | 93.6% | 100.0% | 99.3% | ?Hyperostosis cranalis interna, 144755;Hypermanganesemia with dystonia 2, 617013 |
| SLC39A4 | 100.0% | 100.0% | 100.0% | 99.5% | Acrodermatitis enteropathica, 201100 |
| SLC39A8 | 99.9% | 99.4% | 100.0% | 97.8% | Congenital disorder of glycosylation, type IIn, 616721 |
| SLC3A1 | 96.2% | 96.2% | 100.0% | 99.0% | Cystinuria, 220100 |

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| SLC44A1 | 100.0% | 100.0% | 100.0% | 97.7% | Neurodegeneration, childhood-onset, with ataxia, tremor, optic atrophy, and cognitive decline, 618868 |
| SLC46A1 | 100.0% | 100.0% | 100.0% | 98.7% | Folate malabsorption, hereditary, 229050 |
| SLC52A1 | 100.0% | 100.0% | 100.0% | 99.7% | Riboflavin deficiency, 615026 |
| SLC52A2 | 100.0% | 100.0% | 100.0% | 99.9% | Brown-Vialetto-Van Laere syndrome 2, 614707 |
| SLC52A3 | 100.0% | 100.0% | 100.0% | 99.0% | ?Fazio-Londe disease, 211500;Brown-Vialetto-Van Laere syndrome 1, 211530 |
| SLC5A1 | 100.0% | 100.0% | 100.0% | 98.4% | Glucose/galactose malabsorption, 606824 |
| SLC5A2 | 100.0% | 100.0% | 100.0% | 99.3% | Renal glucosuria, 233100 |
| SLC6A19 | 100.0% | 100.0% | 100.0% | 99.4% | Hartnup disorder, 234500 |
| SLC6A5 | 100.0% | 100.0% | 100.0% | 98.8% | Hyperekplexia 3, 614618 |
| SLC6A6 | 100.0% | 100.0% | 100.0% | 98.4% | Hypotaurinemic retinal degeneration and cardiomyopathy, 145350 |
| SLC6A8 | 100.0% | 99.6% | 95.4% | 67.7% | Cerebral creatine deficiency syndrome 1, 300352 |
| SLC6A9 | 100.0% | 100.0% | 100.0% | 99.5% | Glycine encephalopathy with normal serum glycine, 617301 |
| SLC7A7 | 100.0% | 100.0% | 100.0% | 98.8% | Lysinuric protein intolerance, 222700 |

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| SLC7A9 | 100.0% | 100.0% | 100.0% | 99.0% | Cystinuria, 220100 |
| SLCO1B1 | 100.0% | 100.0% | 100.0% | 97.2% | Hyperbilirubinemia, Rotor type, digenic, 237450 |
| SLCO1B3 | 100.0% | 100.0% | 100.0% | 97.6% | Hyperbilirubinemia, Rotor type, digenic, 237450 |
| SMPD1 | 100.0% | 100.0% | 100.0% | 98.5% | Niemann-Pick disease, type B, 607616;Niemann-Pick disease, type A, 257200 |
| SMS | 100.0% | 99.4% | 97.6% | 72.4% | Intellectual developmental disorder, X-linked syndromic, Snyder-Robinson type, 309583 |
| SNX14 | 95.0% | 95.0% | 100.0% | 98.0% | Spinocerebellar ataxia, autosomal recessive 20, 616354 |
| SOD1 | 100.0% | 100.0% | 100.0% | 99.1% | Spastic tetraplegia and axial hypotonia, progressive, 618598;Amyotrophic lateral sclerosis 1, 105400 |
| SOD2 | 100.0% | 100.0% | 100.0% | 99.4% | {Microvascular complications of diabetes 6}, 612634 |
| SPR | 100.0% | 100.0% | 100.0% | 99.0% | Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716 |
| SPTLC1 | 88.7% | 88.7% | 100.0% | 98.5% | Amyotrophic lateral sclerosis 27, juvenile, 620285;Neuropathy, hereditary sensory and autonomic, type IA, 162400 |

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| SPTLC2 | 100.0% | 100.0% | 100.0% | 98.5% | Neuropathy, hereditary sensory and autonomic, type IC, 613640 |
| SPTSSA | 100.0% | 100.0% | 100.0% | 86.1% | Spastic paraplegia 90A, autosomal dominant, 620416;?Spastic paraplegia 90B, autosomal recessive, 620417 |
| SQOR | 100.0% | 100.0% | 100.0% | 98.2% | Sulfide:quinone oxidoreductase deficiency, 619221 |
| SRD5A2 | 100.0% | 100.0% | 100.0% | 99.3% | Pseudovaginal perineoscrotal hypospadias, 264600 |
| SRD5A3 | 100.0% | 100.0% | 100.0% | 97.6% | Kahrizi syndrome, 612713;Congenital disorder of glycosylation, type Iq, 612379 |
| SSR4 | 100.0% | 99.9% | 97.8% | 72.6% | Congenital disorder of glycosylation, type Iy, 300934 |
| ST3GAL3 | 97.4% | 95.3% | 100.0% | 99.3% | Developmental and epileptic encephalopathy 15, 615006;Intellectual developmental disorder, autosomal recessive 12, 611090 |
| ST3GAL5 | 98.3% | 98.3% | 100.0% | 97.8% | Salt and pepper developmental regression syndrome, 609056 |
| STAR | 100.0% | 100.0% | 100.0% | 99.0% | Lipoid adrenal hyperplasia, 201710 |

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| STS | 96.9% | 96.5% | 98.2% | 72.4% | Ichthyosis, X-linked, 308100 |
| STT3A | 100.0% | 100.0% | 100.0% | 99.0% | Congenital disorder of glycosylation, type Iw, autosomal dominant, 619714;Congenital disorder of glycosylation, type Iw, autosomal recessive, 615596 |
| STT3B | 100.0% | 100.0% | 99.9% | 95.7% | Congenital disorder of glycosylation, type Ix, 615597 |
| STX5 | 100.0% | 100.0% | 100.0% | 98.6% | ?Congenital disorder of glycosylation, type IIaa, 620454 |
| SUCLA2 | 100.0% | 99.6% | 100.0% | 98.8% | Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073 |
| SUCLG1 | 100.0% | 100.0% | 100.0% | 96.4% | Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400 |
| SUCLG2 | 100.0% | 99.8% | 100.0% | 97.2% | |
| SUGCT | 100.0% | 99.9% | 100.0% | 98.7% | Glutaric aciduria III, 231690 |
| SUMF1 | 100.0% | 100.0% | 100.0% | 99.3% | Multiple sulfatase deficiency, 272200 |
| SUOX | 100.0% | 100.0% | 100.0% | 99.0% | Sulfite oxidase deficiency, 272300 |
| TAFAZZIN | 100.0% | 100.0% | 96.7% | 66.1% | Barth syndrome, 302060 |

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| TALDO1 | 100.0% | 100.0% | 100.0% | 98.2% | Transaldolase deficiency, 606003 |
| TANGO2 | 100.0% | 100.0% | 100.0% | 99.4% | Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878 |
| TAT | 100.0% | 100.0% | 100.0% | 98.8% | Tyrosinemia, type II, 276600 |
| TBXAS1 | 100.0% | 100.0% | 100.0% | 98.7% | Ghosal hematodiaphyseal syndrome, 231095 |
| TCIRG1 | 100.0% | 100.0% | 100.0% | 99.6% | Osteopetrosis, autosomal recessive 1, 259700 |
| TCN2 | 94.2% | 94.2% | 100.0% | 98.9% | Transcobalamin II deficiency, 275350 |
| TECR | 100.0% | 100.0% | 100.0% | 99.7% | Intellectual developmental disorder, autosomal recessive 14, 614020 |
| TH | 100.0% | 100.0% | 100.0% | 98.4% | Segawa syndrome, recessive, 605407 |
| TIMM50 | 100.0% | 100.0% | 100.0% | 99.5% | 3-methylglutaconic aciduria, type IX, 617698 |
| TK2 | 100.0% | 100.0% | 100.0% | 98.5% | Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560;?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3, 617069 |

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| TKFC | 100.0% | 100.0% | 100.0% | 99.3% | Triokinase and FMN cyclase deficiency syndrome, 618805 |
| TKT | 98.1% | 98.1% | 100.0% | 99.0% | Short stature, developmental delay, and congenital heart defects, 617044 |
| TMEM106B | 100.0% | 100.0% | 100.0% | 98.4% | Leukodystrophy, hypomyelinating, 16, 617964 |
| TMEM165 | 100.0% | 100.0% | 100.0% | 97.8% | Congenital disorder of glycosylation, type IIk, 614727 |
| TMEM199 | 100.0% | 100.0% | 100.0% | 98.7% | Congenital disorder of glycosylation, type IIp, 616829 |
| TMEM70 | 100.0% | 100.0% | 100.0% | 97.4% | Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052 |
| TMLHE | 99.9% | 99.4% | 98.1% | 78.7% | {Autism, susceptibility to, X-linked 6}, 300872 |
| TNIK | 100.0% | 100.0% | 100.0% | 98.5% | Intellectual developmental disorder, autosomal recessive 54, 617028 |
| TPI1 | 100.0% | 100.0% | 100.0% | 98.0% | Hemolytic anemia due to triosephosphate isomerase deficiency, 615512 |
| TPK1 | 100.0% | 100.0% | 100.0% | 98.0% | Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458 |

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| TPMT | 100.0% | 100.0% | 100.0% | 98.1% | {Thiopurines, poor metabolism of, 1}, 610460 |
| TPP1 | 100.0% | 100.0% | 100.0% | 99.2% | Ceroid lipofuscinosis, neuronal, 2, 204500;Spinocerebellar ataxia, autosomal recessive 7, 609270 |
| TRAK1 | 100.0% | 100.0% | 100.0% | 99.1% | Developmental and epileptic encephalopathy 68, 618201 |
| TRAPPC11 | 100.0% | 100.0% | 100.0% | 98.3% | Muscular dystrophy, limb-girdle, autosomal recessive 18, 615356 |
| TRAPPC2L | 100.0% | 100.0% | 100.0% | 99.8% | Encephalopathy, progressive, early-onset, with episodic rhabdomyolysis, 618331 |
| TRAPPC9 | 100.0% | 100.0% | 100.0% | 98.7% | Intellectual developmental disorder, autosomal recessive 13, 613192 |
| TREH | 100.0% | 100.0% | 100.0% | 99.4% | Trehalase deficiency, 612119 |
| TUSC3 | 100.0% | 100.0% | 100.0% | 98.4% | Intellectual developmental disorder, autosomal recessive 7, 611093 |
| TYMP | 100.0% | 100.0% | 100.0% | 98.6% | Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041 |
| TYMS | 100.0% | 100.0% | 100.0% | 96.4% | Dyskeratosis congenita, digenic, 620040 |

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| TYR | 100.0% | 99.9% | 100.0% | 98.8% | [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 | 100.0% | 100.0% | 100.0% | 98.9% | [Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)], 612271;Albinism, oculocutaneous, type III, 203290 |
| UFM1 | 100.0% | 100.0% | 100.0% | 99.0% | Leukodystrophy, hypomyelinating, 14, 617899 |
| UGT1A1 | 100.0% | 100.0% | 100.0% | 98.8% | 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 | 100.0% | 100.0% | 100.0% | 99.4% | Orotic aciduria, 258900 |
| UPB1 | 100.0% | 100.0% | 100.0% | 98.8% | Beta-ureidopropionase deficiency, 613161 |

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| UROC1 | 100.0% | 100.0% | 100.0% | 99.5% | ?Urocanase deficiency, 276880 |
| UROD | 100.0% | 100.0% | 100.0% | 99.2% | Porphyria, hepatoerythropoietic, 176100;Porphyria cutanea tarda, 176100 |
| UROS | 100.0% | 100.0% | 100.0% | 98.2% | Porphyria, congenital erythropoietic, 263700 |
| VMA21 | 100.0% | 100.0% | 98.5% | 72.2% | Myopathy, X-linked, with excessive autophagy, 310440 |
| VPS13B | 100.0% | 99.8% | 100.0% | 98.7% | Cohen syndrome, 216550 |
| VPS33A | 89.5% | 89.5% | 100.0% | 96.4% | Mucopolysaccharidosis-plus syndrome, 617303 |
| XDH | 100.0% | 100.0% | 100.0% | 99.2% | Xanthinuria, type I, 278300 |
| XYLT1 | 100.0% | 99.8% | 99.6% | 93.5% | Desbuquois dysplasia 2, 615777;{Pseudoxanthoma elasticum, modifier of severity of}, 264800 |
| XYLT2 | 99.9% | 99.2% | 100.0% | 98.9% | {Pseudoxanthoma elasticum, modifier of severity of}, 264800;Spondyloocular syndrome, 605822 |
| ZBTB11 | 100.0% | 100.0% | 100.0% | 99.1% | Intellectual developmental disorder, autosomal recessive 69, 618383 |
| ZMPSTE24 | 100.0% | 100.0% | 100.0% | 98.7% | Mandibuloacral dysplasia with type B lipodystrophy, 608612;Restrictive dermopathy 1, 275210 |

*Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan 43(Database issue):D1079-85.*
*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.*
*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.*
*srWGS GRCh38 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 GRCh38 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 : March 17th, 2023.*
*This list is accurate for panel version DG 4.0.0*

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