

# METABOLIC DISORDERS PANEL DG-5.1.0 (827 GENES)

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

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

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

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

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| AK1      | 100%  | 100%  | 100% | 100%  | 99.8% | Anemia, congenital, nonspherocytic hemolytic, 3, adenylate kinase deficient, 612631                                                                                                              |
| AK2      | 98.1% | 93.2% | 100% | 100%  | 99.1% | Reticular dysgenesis, 267500                                                                                                                                                                     |
| AKR1C1   | 100%  | 100%  | 100% | 100%  | 99.5% |                                                                                                                                                                                                  |
| AKR1D1   | 88.4% | 88.4% | 100% | 100%  | 99.8% | Bile acid synthesis defect, congenital, 2, 235555                                                                                                                                                |
| ALAD     | 100%  | 100%  | 100% | 100%  | 99.4% | Porphyria, acute hepatic, 612740;{Lead poisoning, susceptibility to}, 612740                                                                                                                     |
| ALAS2    | 100%  | 100%  | 99%  | 90.9% | 70.9% | Anemia, sideroblastic, 1, 300751;Protoporphyria, erythropoietic, X-linked, 300752                                                                                                                |
| ALDH18A1 | 100%  | 100%  | 100% | 100%  | 99.6% | Spastic paraplegia 9A, autosomal dominant, 601162;Cutis laxa, autosomal recessive, type IIIA, 219150;Spastic paraplegia 9B, autosomal recessive, 616586;Cutis laxa, autosomal dominant 3, 616603 |
| ALDH1A3  | 94.4% | 94.4% | 100% | 99.9% | 99.4% | Microphthalmia, isolated 8, 615113                                                                                                                                                               |
| ALDH2    | 100%  | 99.9% | 100% | 100%  | 99.4% | {Sublingual nitroglycerin, susceptibility to poor response to}, 610251;Alcohol sensitivity, acute, 610251;{Hangover, susceptibility to}, 610251                                                  |
| ALDH3A2  | 93.5% | 93.5% | 100% | 100%  | 99.7% | Sjogren-Larsson syndrome, 270200                                                                                                                                                                 |

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| ALDH4A1 | 97.9% | 96.1% | 100%  | 100%  | 99.5% | Hyperprolinemia, type II, 239510                                                                                                                                                                                                             |
| ALDH5A1 | 100%  | 100%  | 100%  | 100%  | 99.5% | Succinic semialdehyde dehydrogenase deficiency, 271980                                                                                                                                                                                       |
| ALDH6A1 | 95.2% | 95.2% | 100%  | 100%  | 99.7% | Methylmalonate semialdehyde dehydrogenase deficiency, 614105                                                                                                                                                                                 |
| ALDH7A1 | 97.2% | 97.2% | 100%  | 100%  | 99.8% | Epilepsy, early-onset, 4, vitamin B6-dependent, 266100                                                                                                                                                                                       |
| ALDOA   | 100%  | 100%  | 100%  | 100%  | 99.4% | Glycogen storage disease XII, 611881                                                                                                                                                                                                         |
| ALDOB   | 100%  | 100%  | 100%  | 99.9% | 99.8% | Fructose intolerance, hereditary, 229600                                                                                                                                                                                                     |
| ALG1    | 100%  | 100%  | 100%  | 100%  | 99.7% | Congenital disorder of glycosylation, type Ik, 608540                                                                                                                                                                                        |
| ALG10   | 100%  | 100%  | 100%  | 100%  | 99.8% |                                                                                                                                                                                                                                              |
| ALG11   | 91%   | 91%   | 100%  | 100%  | 99.9% | Congenital disorder of glycosylation, type Ip, 613661                                                                                                                                                                                        |
| ALG12   | 100%  | 100%  | 100%  | 100%  | 99.6% | Congenital disorder of glycosylation, type Ig, 607143                                                                                                                                                                                        |
| ALG13   | 100%  | 100%  | 99.2% | 90.7% | 72.3% | Developmental and epileptic encephalopathy 36, 300884                                                                                                                                                                                        |
| ALG14   | 100%  | 100%  | 100%  | 100%  | 99.8% | Intellectual developmental disorder with epilepsy, behavioral abnormalities, and coarse facies, 619031;Myopathy, epilepsy, and progressive cerebral atrophy, 619036;?Myasthenic syndrome, congenital, 15, without tubular aggregates, 616227 |

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

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

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

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

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

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

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

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

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

|       |       |       |      |       |       |                                                                                                                                                                                                        |
|-------|-------|-------|------|-------|-------|--------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| CIAO1 | 100%  | 100%  | 100% | 99.7% | 98.8% | Multiple mitochondrial dysfunctions syndrome 10, 620960                                                                                                                                                |
| CIDEC | 100%  | 100%  | 100% | 100%  | 99.8% | ?Lipodystrophy, familial partial, type 5, 615238                                                                                                                                                       |
| CLCN7 | 97.5% | 97.5% | 100% | 100%  | 99.7% | Hypopigmentation, organomegaly, and delayed myelination and development, 618541;Osteopetrosis, autosomal recessive 4, 611490;Osteopetrosis, autosomal dominant 2, 166600                               |
| CLN3  | 93.1% | 93.1% | 100% | 100%  | 99.4% | Ceroid lipofuscinosis, neuronal, 3, 204200;Retinitis pigmentosa 101, 621548                                                                                                                            |
| CLN5  | 83%   | 83%   | 100% | 100%  | 99.8% | Ceroid lipofuscinosis, neuronal, 5, 256731                                                                                                                                                             |
| CLN6  | 95.1% | 95.1% | 100% | 100%  | 99.4% | Ceroid lipofuscinosis, neuronal, 6B (Kufs type), 204300;Ceroid lipofuscinosis, neuronal, 6A, 601780                                                                                                    |
| CLN8  | 100%  | 100%  | 100% | 100%  | 99.5% | Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003;Ceroid lipofuscinosis, neuronal, 8, 600143                                                                                       |
| CLPB  | 98.2% | 98.2% | 100% | 100%  | 99.7% | Neutropenia, severe congenital, 9, autosomal dominant, 619813;3-methylglutac onic aciduria, type VIIB, autosomal recessive, 616271;3-methylglutac onic aciduria, type VIIA, autosomal dominant, 619835 |
| CMAS  | 100%  | 100%  | 100% | 100%  | 99.9% |                                                                                                                                                                                                        |
| COG1  | 100%  | 100%  | 100% | 100%  | 99.4% | Congenital disorder of glycosylation, type IIg, 611209                                                                                                                                                 |

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

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

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

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

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

|        |       |       |      |       |       |                                                                                                                                                                                                                        |
|--------|-------|-------|------|-------|-------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| DDC    | 100%  | 100%  | 100% | 100%  | 99.7% | Aromatic L-amino acid decarboxylase deficiency, 608643                                                                                                                                                                 |
| DDHD1  | 100%  | 100%  | 100% | 100%  | 99.3% | Spastic paraplegia 28, autosomal recessive, 609340                                                                                                                                                                     |
| DDOST  | 100%  | 100%  | 100% | 99.9% | 99.2% | Congenital disorder of glycosylation, type I <sub>r</sub> , 614507                                                                                                                                                     |
| DEGS1  | 100%  | 100%  | 100% | 100%  | 99.8% | Leukodystrophy, hypomyelinating, 18, 618404                                                                                                                                                                            |
| DGAT1  | 100%  | 100%  | 100% | 100%  | 99.6% | Diarrhea 7, protein-losing enteropathy type, 615863                                                                                                                                                                    |
| DGKE   | 100%  | 100%  | 100% | 100%  | 99.7% | {Hemolytic uremic syndrome, atypical, susceptibility to, 7}, 615008;Nephrotic syndrome, type 7, 615008                                                                                                                 |
| DGUOK  | 100%  | 100%  | 100% | 100%  | 99.9% | Portal hypertension, noncirrhotic, 1, 617068;Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, 617070;Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880 |
| DHCR24 | 97.8% | 97.8% | 100% | 99.7% | 98.6% | Desmosterolosis, 602398                                                                                                                                                                                                |
| DHCR7  | 96.2% | 96.2% | 100% | 100%  | 99.6% | Smith-Lemli-Opitz syndrome, 270400                                                                                                                                                                                     |

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

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

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

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

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| EXTL3  | 100%  | 99.9% | 100% | 100% | 99.7% | Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425                                                                                                                                                                                      |
| EYA1   | 100%  | 100%  | 100% | 100% | 100%  | Branchioototic syndrome 1, 602588;Branchiootorenal syndrome 1, with or without cataracts, 113650;Anterior segment anomalies with or without cataract, 602588;?Otofaciocervical syndrome, 166780                                                             |
| FA2H   | 100%  | 100%  | 100% | 100% | 99.7% | Spastic paraplegia 35, autosomal recessive, 612319                                                                                                                                                                                                          |
| FAH    | 95.2% | 93.3% | 100% | 100% | 99.5% | Tyrosinemia, type I, 276700                                                                                                                                                                                                                                 |
| FAM20B | 100%  | 100%  | 100% | 100% | 99.9% |                                                                                                                                                                                                                                                             |
| FAR1   | 91%   | 90.9% | 100% | 100% | 99.9% | Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154;Cataracts, spastic paraparesis, and speech delay, 619338                                                                                                                                            |
| FBN1   | 100%  | 100%  | 100% | 100% | 99.8% | Geleophysic dysplasia 2, 614185;Weill-Marchesani syndrome 2, dominant, 608328;Ectopia lentis, familial, 129600;MASS syndrome, 604308;Marfan lipodystrophy syndrome, 616914;Acromicric dysplasia, 102370;Marfan syndrome, 154700;Stiff skin syndrome, 184900 |
| FBP1   | 95.5% | 94.8% | 100% | 100% | 99.7% | Fructose-1,6-bisphosphatase deficiency, 229700                                                                                                                                                                                                              |

|       |      |       |      |      |       |                                                                                                                                                                                                                                                                                                   |
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| FBP2  | 100% | 100%  | 100% | 100% | 99.7% | ?Leukodystrophy, childhood-onset, remitting, 619864                                                                                                                                                                                                                                               |
| FCSK  | 100% | 100%  | 100% | 100% | 99.6% | Congenital disorder of glycosylation with defective fucosylation 2, 618324                                                                                                                                                                                                                        |
| FDFT1 | 100% | 98.7% | 100% | 100% | 99.5% | Squalene synthase deficiency, 618156                                                                                                                                                                                                                                                              |
| FECH  | 100% | 100%  | 100% | 100% | 99.8% | Protoporphyrria, erythropoietic, 1, 177000                                                                                                                                                                                                                                                        |
| FH    | 100% | 100%  | 100% | 100% | 99.8% | Leiomyomatosis and renal cell cancer, 150800;Fumarase deficiency, 606812                                                                                                                                                                                                                          |
| FKRP  | 100% | 100%  | 100% | 100% | 99.3% | Muscular dystrophy-dystroglycanopathy (congenital with or without impaired intellectual development), type B, 5, 606612;Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153 |

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

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

|         |       |       |      |       |       |                                                                                                                                                                                                                                                                                            |
|---------|-------|-------|------|-------|-------|--------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| GALNT14 | 100%  | 100%  | 100% | 99.9% | 99.4% |                                                                                                                                                                                                                                                                                            |
| GALNT2  | 100%  | 100%  | 100% | 100%  | 99.4% | Congenital disorder of glycosylation, type II, 618885                                                                                                                                                                                                                                      |
| GALNT3  | 100%  | 100%  | 100% | 100%  | 99.8% | Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900                                                                                                                                                                                                                                 |
| GALT    | 100%  | 100%  | 100% | 100%  | 99.5% | Galactosemia, 230400                                                                                                                                                                                                                                                                       |
| GAMT    | 97%   | 93.2% | 100% | 99.8% | 98.8% | Cerebral creatine deficiency syndrome 2, 612736                                                                                                                                                                                                                                            |
| GANAB   | 100%  | 100%  | 100% | 100%  | 99.6% | Polycystic kidney disease 3, 600666                                                                                                                                                                                                                                                        |
| GATM    | 100%  | 100%  | 100% | 100%  | 99.9% | Cerebral creatine deficiency syndrome 3, 612718;Fanconi renotubular syndrome 1, 134600                                                                                                                                                                                                     |
| GBA1    | 100%  | 98.3% | 100% | 100%  | 99.5% | {Lewy body dementia, susceptibility to}, 127750;Gaucher disease, type II, 230900;Gaucher disease, type IIIC, 231005;Gaucher disease, type III, 231000;Gaucher disease, type I, 230800;Gaucher disease, perinatal lethal, 608013;{Parkinson disease, late-onset, susceptibility to}, 168600 |
| GBA2    | 100%  | 100%  | 100% | 99.9% | 99.4% | Spastic paraplegia 46, autosomal recessive, 614409                                                                                                                                                                                                                                         |
| GBE1    | 100%  | 100%  | 100% | 100%  | 99.7% | Glycogen storage disease IV, 232500;Polyglucosan body disease, adult form, 263570                                                                                                                                                                                                          |
| GCDH    | 79.8% | 79.8% | 100% | 99.9% | 99.5% | Glutaricaciduria, type I, 231670                                                                                                                                                                                                                                                           |

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

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

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

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

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

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

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

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

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

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

|        |       |       |       |       |       |                                                                                                                                                                                                                                      |
|--------|-------|-------|-------|-------|-------|--------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| KCNJ11 | 100%  | 100%  | 100%  | 100%  | 99.5% | Diabetes, permanent neonatal 2, with or without neurologic features, 618856;Maturity-onset diabetes of the young, type 13, 616329;Diabetes mellitus, transient neonatal 3, 610582;Hyperinsulinemic hypoglycemia, familial, 2, 601820 |
| KGD4   | 100%  | 100%  | 100%  | 99.8% | 99.5% |                                                                                                                                                                                                                                      |
| KMT2A  | 99.2% | 99.2% | 100%  | 100%  | 99.5% | Wiedemann-Steiner syndrome, 605130                                                                                                                                                                                                   |
| KMT2D  | 100%  | 100%  | 100%  | 100%  | 99.2% | Branchial arch abnormalities, choanal atresia, athelia, hearing loss, and hypothyroidism syndrome, 620186;Kabuki syndrome 1, 147920                                                                                                  |
| L2HGDH | 92.8% | 92.8% | 100%  | 100%  | 99.6% | L-2-hydroxyglutaric aciduria, 236792                                                                                                                                                                                                 |
| LAMP2  | 70.7% | 70.7% | 98.9% | 91%   | 71.6% | Danon disease, 300257                                                                                                                                                                                                                |
| LARGE1 | 96.9% | 96.9% | 100%  | 100%  | 99.6% | Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 6, 608840;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154                      |
| LCAT   | 100%  | 100%  | 100%  | 99.9% | 99%   | Fish-eye disease, 136120;Norum disease, 245900                                                                                                                                                                                       |
| LCT    | 100%  | 100%  | 100%  | 100%  | 99.5% | Lactase deficiency, congenital, 223000                                                                                                                                                                                               |
| LDHA   | 100%  | 100%  | 100%  | 100%  | 99.6% | Glycogen storage disease XI, 612933                                                                                                                                                                                                  |

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

|       |       |       |      |       |       |                                                                                                                                                                                                                                                                                                                                                                                                                                                                                     |
|-------|-------|-------|------|-------|-------|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| LMNA  | 100%  | 100%  | 100% | 99.9% | 99.3% | Mandibuloacral dysplasia, 248370;Heart-hand syndrome, Slovenian type, 610140;Cardiomyopathy, dilated, 1A, 115200;Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516;Restrictive dermopathy 2, 619793;Charcot-Marie-Tooth disease, type 2B1, 605588;Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350;Hutchinson-Gilford progeria, 176670;Lipodystrophy, familial partial, type 2, 151660;Muscular dystrophy, congenital, 613205;Malouf syndrome, 212112 |
| LMNB2 | 92.9% | 92.9% | 100% | 99.8% | 98.3% | Microcephaly 27, primary, autosomal dominant, 619180;?Epilepsy, progressive myoclonic, 9, 616540;{Lipodystrophy, partial, acquired, susceptibility to}, 608709                                                                                                                                                                                                                                                                                                                      |
| LPIN1 | 94%   | 94%   | 100% | 100%  | 99.7% | Myoglobinuria, acute recurrent, autosomal recessive, 268200                                                                                                                                                                                                                                                                                                                                                                                                                         |
| LPIN2 | 99.3% | 99.2% | 100% | 99.9% | 99.4% | Majeed syndrome, 609628                                                                                                                                                                                                                                                                                                                                                                                                                                                             |
| LPL   | 100%  | 100%  | 100% | 100%  | 99.6% | Lipoprotein lipase deficiency, 238600;[High density lipoprotein cholesterol level QTL 11], 238600;Combined hyperlipidemia, familial, 144250                                                                                                                                                                                                                                                                                                                                         |

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

|         |      |       |      |      |       |                                                                                                                                                                                                                                                                                                                              |
|---------|------|-------|------|------|-------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| POMGNT1 | 100% | 100%  | 100% | 100% | 99.6% | Muscular dystrophy-dystroglycan opathy (limb-girdle), type C, 3, 613157; Muscular dystrophy-dystroglycan opathy (congenital with impaired intellectual development), type B, 3, 613151; Retinitis pigmentosa 76, 617123; Muscular dystrophy-dystroglycan opathy (congenital with brain and eye anomalies), type A, 3, 253280 |
| POMGNT2 | 100% | 100%  | 100% | 100% | 99.5% | Muscular dystrophy-dystroglycan opathy (congenital with brain and eye anomalies), type A, 8, 614830; Muscular dystrophy-dystroglycan opathy (limb-girdle) type C, 8, 618135                                                                                                                                                  |
| POMK    | 100% | 100%  | 100% | 100% | 99.8% | ?Muscular dystrophy-dystroglycan opathy (limb-girdle), type C, 12, 616094; Muscular dystrophy-dystroglycan opathy (congenital with brain and eye anomalies), type A, 12, 615249                                                                                                                                              |
| POMT1   | 100% | 99.7% | 100% | 100% | 99.3% | Muscular dystrophy-dystroglycan opathy (congenital with brain and eye anomalies), type A, 1, 236670; Muscular dystrophy-dystroglycan opathy (limb-girdle), type C, 1, 609308; Muscular dystrophy-dystroglycan opathy (congenital with impaired intellectual development), type B, 1, 613155                                  |

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

Gene symbols used follow HGNC guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan 43(Database issue):D1079-85.

*TWIST X2 covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.*

*TWIST X2 covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.*

*srWGS covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WGS mapped against GRCh38.*

*srWGS covered 15x describes the percentage of a gene's coding sequence that is covered at least 15x when analyzed by WGS mapped against GRCh38.*

*srWGS covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WGS mapped against GRCh38.*

*non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.*

*OMIM release used for OMIM disease identifiers and descriptions : November 25th, 2024.*

*This list is accurate for panel version DG 5.1.0*

*Ad 1. Blank field signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors*