# METABOLIC DISORDERS GENE PANEL DG 2.18 (683 genes)

Releasedate: 20-04-2020

| Gene | Agilent V5 covered >10x | Agilent V5 covered > 20x | TWIST covered >10x | TWIST covered >20x | Associated Phenotype description and OMIM disease ID |
|---|---|---|---|---|---|
| *AASS* | 100% | 99,70% | 100% | 100% | Hyperlysinemia, 238700 |
| *ABAT* | 100% | 99,40% | 100% | 100% | GABA-transaminase deficiency, 613163 |
| *ABCC8* | 100% | 99,80% | 100% | 100% | Diabetes mellitus, permanent neonatal, 606176<br>Diabetes mellitus, noninsulin-dependent, 125853<br>Diabetes mellitus, transient neonatal 2, 610374<br>Hyperinsulinemic hypoglycemia, familial, 1, 256450<br>Hypoglycemia of infancy, leucine-sensitive, 240800 |
| *ABCD1* | 75,80% | 71,60% | 100% | 100% | Adrenomyeloneuropathy, adult, 300100<br>Adrenoleukodystrophy, 300100 |
| *ABCD2* | 100% | 99,80% | 100% | 100% | No OMIM disease ID |
| *ABCD3* | 99,80% | 97,70% | 100% | 100% | ?Bile acid synthesis defect, congenital, 5, 616278 |
| *ABCD4* | 99,90% | 98,60% | 100% | 100% | Methylmalonic aciduria and homocystinuria, cblJ type, 614857 |
| *ABCG5* | 100% | 100% | 100% | 100% | Sitosterolemia 2, 618666 |
| *ABCG8* | 99,10% | 97,30% | 100% | 100% | Sitosterolemia 1, 210250 |
| *ABHD12* | 98,70% | 92,30% | 100% | 99,30% | Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674 |
| *ABHD5* | 100% | 100% | 100% | 100% | Chanarin-Dorfman syndrome, 275630 |
| *ACACA* | 98,40% | 98,10% | 100% | 100% | No OMIM disease ID |
| *ACAD8* | 100% | 100% | 100% | 100% | Isobutyryl-CoA dehydrogenase deficiency, 611283 |
| *ACAD9* | 100% | 99,90% | 100% | 100% | Mitochondrial complex I deficiency, nuclear type 20, 611126 |
| *ACADM* | 99,80% | 99,00% | 100% | 100% | Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450 |
| *ACADS* | 99,90% | 98,20% | 100% | 100% | Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470 |
| *ACADSB* | 100% | 99,20% | 100% | 100% | 2-methylbutyrylglycinuria, 610006 |
| *ACADVL* | 99,40% | 97,30% | 100% | 100% | VLCAD deficiency, 201475 |
| *ACAT1* | 99,90% | 97,50% | 100% | 100% | Alpha-methylacetoacetic aciduria, 203750 |
| *ACAT2* | 100% | 100% | 100% | 100% | No OMIM disease ID |
| *ACBD5* | 100% | 99,20% | 100% | 100% | Retinal dystrophy with leukodystrophy, 618863 |
| *ACO2* | 96,30% | 90,30% | 100% | 100% | ?Optic atrophy 9, 616289<br>Infantile cerebellar-retinal degeneration, 614559 |
| *ACOX1* | 100% | 99,90% | 100% | 100% | Peroxisomal acyl-CoA oxidase deficiency, 264470 |

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| *ACOX2* | 100% | 99,20% | 100% | 100% | Bile acid synthesis defect, congenital, 6, 617308 |
| *ACSF3* | 100% | 99,90% | 100% | 100% | Combined malonic and methylmalonic aciduria, 614265 |
| *ACSL4* | 98,70% | 94,60% | 100% | 100% | Mental retardation, X-linked 63, 300387 |
| *ACY1* | 100% | 98,80% | 100% | 100% | Aminoacylase 1 deficiency, 609924 |
| *ADA* | 100% | 99,70% | 100% | 100% | Adenosine deaminase deficiency, partial, 102700<br>Severe combined immunodeficiency due to ADA deficiency, 102700 |
| *ADCK5* | 100% | 99,90% | 100% | 100% | No OMIM disease ID |
| *ADCY5* | 95,10% | 91,20% | 99,20% | 98,00% | Dyskinesia, familial, with facial myokymia, 606703 |
| *ADK* | 99,50% | 95,80% | 100% | 100% | Hypermethioninemia due to adenosine kinase deficiency, 614300 |
| *ADSL* | 99,20% | 98,70% | 100% | 100% | Adenylosuccinase deficiency, 103050 |
| *AGA* | 100% | 100% | 100% | 100% | Aspartylglucosaminuria, 208400 |
| *AGK* | 99,90% | 97,60% | 100% | 100% | Sengers syndrome, 212350<br>Cataract 38, autosomal recessive, 614691 |
| *AGL* | 100% | 99,40% | 100% | 100% | Glycogen storage disease IIIb, 232400<br>Glycogen storage disease IIIa, 232400 |
| *AGPAT2* | 99,60% | 96,10% | 100% | 100% | Lipodystrophy, congenital generalized, type 1, 608594 |
| *AGPS* | 99,30% | 95,40% | 100% | 99,90% | Rhizomelic chondrodysplasia punctata, type 3, 600121 |
| *AGXT* | 100% | 100% | 100% | 100% | Hyperoxaluria, primary, type 1, 259900 |
| *AHCY* | 100% | 99,20% | 100% | 100% | Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752 |
| *AK1* | 100% | 100% | 100% | 100% | Hemolytic anemia due to adenylate kinase deficiency, 612631 |
| *AK2* | 98,80% | 94,50% | 100% | 100% | Reticular dysgenesis, 267500 |
| *AKR1D1* | 100% | 99,40% | 100% | 100% | Bile acid synthesis defect, congenital, 2, 235555 |
| *ALAD* | 99,30% | 94,10% | 100% | 100% | Porphyria, acute hepatic, 612740 |
| *ALAS2* | 98,90% | 94,90% | 100% | 100% | Protoporphyria, erythropoietic, X-linked, 300752<br>Anemia, sideroblastic, 1, 300751 |
| *ALDH18A1* | 100% | 99,90% | 100% | 100% | Cutis laxa, autosomal recessive, type IIIA, 219150<br>Cutis laxa, autosomal dominant 3, 616603<br>Spastic paraplegia 9B, autosomal recessive, 616586<br>Spastic paraplegia 9A, autosomal dominant, 601162 |
| *ALDH1A3* | 97,20% | 94,50% | 100% | 99,90% | Microphthalmia, isolated 8, 615113 |
| *ALDH2* | 100% | 100% | 100% | 100% | Alcohol sensitivity, acute, 610251 |
| *ALDH3A2* | 95,30% | 94,60% | 100% | 100% | Sjogren-Larsson syndrome, 270200 |
| *ALDH4A1* | 100% | 99,40% | 100% | 100% | Hyperprolinemia, type II, 239510 |
| *ALDH5A1* | 91,00% | 81,50% | 100% | 100% | Succinic semialdehyde dehydrogenase deficiency, 271980 |
| *ALDH6A1* | 100% | 99,90% | 100% | 100% | Methylmalonate semialdehyde dehydrogenase deficiency, 614105 |
| *ALDH7A1* | 94,40% | 88,80% | 100% | 100% | Epilepsy, pyridoxine-dependent, 266100 |
| *ALDOA* | 75,50% | 74,00% | 100% | 100% | Glycogen storage disease XII, 611881 |

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| *ALDOB* | 100% | 99,10% | 100% | 100% | Fructose intolerance, hereditary, 229600 |
| *ALG1* | 53,00% | 45,80% | 100% | 100% | Congenital disorder of glycosylation, type Ik, 608540 |
| *ALG10* | 100% | 100% | 100% | 100% | No OMIM disease ID |
| *ALG11* | 96,80% | 96,80% | 96,80% | 96,80% | Congenital disorder of glycosylation, type Ip, 613661 |
| *ALG12* | 100% | 100% | 100% | 100% | Congenital disorder of glycosylation, type Ig, 607143 |
| *ALG13* | 98,40% | 92,60% | 100% | 99,60% | Epileptic encephalopathy, early infantile, 36, 300884<br>?Congenital disorder of glycosylation, type Is, 300884 |
| *ALG14* | 100% | 99,90% | 100% | 100% | ?Myasthenic syndrome, congenital, 15, without tubular aggregates, 616227 |
| *ALG2* | 100% | 100% | 100% | 100% | Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228<br>?Congenital disorder of glycosylation, type Ii, 607906 |
| *ALG3* | 100% | 99,70% | 100% | 100% | Congenital disorder of glycosylation, type Id, 601110 |
| *ALG6* | 98,60% | 94,80% | 100% | 100% | Congenital disorder of glycosylation, type Ic, 603147 |
| *ALG8* | 97,20% | 95,60% | 96,60% | 96,60% | Congenital disorder of glycosylation, type Ih, 608104<br>Polycystic liver disease 3 with or without kidney cysts, 617874 |
| *ALG9* | 100% | 99,70% | 100% | 100% | Gillessen-Kaesbach-Nishimura syndrome, 263210<br>Congenital disorder of glycosylation, type Il, 608776 |
| *ALOX12B* | 100% | 100% | 100% | 100% | Ichthyosis, congenital, autosomal recessive 2, 242100 |
| *ALPL* | 100% | 100% | 100% | 100% | Hypophosphatasia, adult, 146300<br>Odontohypophosphatasia, 146300<br>Hypophosphatasia, childhood, 241510<br>Hypophosphatasia, infantile, 241500 |
| *AMACR* | 100% | 100% | 100% | 100% | Bile acid synthesis defect, congenital, 4, 214950<br>Alpha-methylacyl-CoA racemase deficiency, 614307 |
| *AMN* | 89,70% | 80,00% | 100% | 100% | Megaloblastic anemia-1, Norwegian type, 261100 |
| *AMPD1* | 99,90% | 98,60% | 100% | 100% | Myopathy due to myoadenylate deaminase deficiency, 615511 |
| *AMPD3* | 99,90% | 98,50% | 100% | 100% | No OMIM disease ID |
| *AMT* | 100% | 100% | 100% | 100% | Glycine encephalopathy, 605899 |
| *AP1S1* | 99,90% | 99,50% | 100% | 100% | MEDNIK syndrome, 609313 |
| *AP3B2* | 99,40% | 95,10% | 100% | 100% | Epileptic encephalopathy, early infantile, 48, 617276 |
| *APOC2* | 100% | 100% | 100% | 100% | Hyperlipoproteinemia, type Ib, 207750 |
| *APRT* | 100% | 99,50% | 100% | 100% | Adenine phosphoribosyltransferase deficiency, 614723 |
| *ARG1* | 100% | 100% | 100% | 100% | Argininemia, 207800 |
| *ARSA* | 100% | 99,80% | 100% | 100% | Metachromatic leukodystrophy, 250100 |
| *ARSB* | 96,90% | 88,30% | 100% | 100% | Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200 |
| *ASAH1* | 99,70% | 98,60% | 100% | 100% | Farber lipogranulomatosis, 228000<br>Spinal muscular atrophy with progressive myoclonic epilepsy, 159950 |
| *ASL* | 100% | 99,60% | 100% | 100% | Argininosuccinic aciduria, 207900 |

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| *ASNS* | 99,40% | 95,20% | 100% | 100% | Asparagine synthetase deficiency, 615574 |
| *ASPA* | 99,90% | 98,30% | 100% | 100% | Canavan disease, 271900 |
| *ASS1* | 95,40% | 87,90% | 100% | 100% | Citrullinemia, 215700 |
| *ATIC* | 99,90% | 99,30% | 100% | 100% | AICA-ribosiduria due to ATIC deficiency, 608688 |
| *ATP1A1* | 100% | 100% | 100% | 100% | Charcot-Marie-Tooth disease, axonal, type 2DD, 618036<br>Hypomagnesemia, seizures, and mental retardation 2, 618314 |
| *ATP6AP1* | 98,20% | 92,10% | 100% | 100% | Immunodeficiency 47, 300972 |
| *ATP6V0A2* | 100% | 99,50% | 100% | 100% | Wrinkly skin syndrome, 278250<br>Cutis laxa, autosomal recessive, type IIA, 219200 |
| *ATP6V1A* | 99,90% | 98,70% | 100% | 100% | Epileptic encephalopathy, infantile or early childhood, 3, 618012<br>Cutis laxa, autosomal recessive, type IID, 617403 |
| *ATP6V1E1* | 93,10% | 88,30% | 100% | 100% | Cutis laxa, autosomal recessive, type IIC, 617402 |
| *ATP7A* | 99,70% | 97,50% | 100% | 100% | Occipital horn syndrome, 304150<br>Menkes disease, 309400<br>Spinal muscular atrophy, distal, X-linked 3, 300489 |
| *ATP7B* | 99,90% | 99,20% | 100% | 100% | Wilson disease, 277900 |
| *ATP8B1* | 96,50% | 94,00% | 100% | 100% | Cholestasis, intrahepatic, of pregnancy, 1, 147480<br>Cholestasis, progressive familial intrahepatic 1, 211600<br>Cholestasis, benign recurrent intrahepatic, 243300 |
| *AUH* | 100% | 99,80% | 100% | 100% | 3-methylglutaconic aciduria, type I, 250950 |
| *B3GALNT1* | 100% | 99,80% | 100% | 100% | No OMIM disease ID |
| *B3GALNT2* | 93,80% | 89,40% | 92,50% | 92,50% | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181 |
| *B3GALT6* | 75,70% | 69,70% | 89,80% | 81,60% | Al-Gazali syndrome, 609465<br>Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349<br>Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640 |
| *B3GAT3* | 99,90% | 98,20% | 94,80% | 94,80% | Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600 |
| *B3GLCT* | 99,60% | 96,30% | 99,90% | 99,20% | Peters-plus syndrome, 261540 |
| *B4GALT1* | 100% | 99,80% | 100% | 100% | Congenital disorder of glycosylation, type IId, 607091 |
| *B4GALT7* | 99,80% | 97,40% | 99,90% | 98,60% | Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070 |
| *B4GAT1* | 100% | 100% | 100% | 100% | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287 |
| *BAAT* | 99,80% | 98,40% | 100% | 100% | Hypercholanemia, familial, 607748 |
| *BCAT1* | 100% | 100% | 100% | 100% | No OMIM disease ID |
| *BCAT2* | 100% | 100% | 100% | 100% | ?Hypervalinemia or hyperleucine-isoleucinemia, 618850 |
| *BCKDHA* | 99,90% | 99,20% | 100% | 100% | Maple syrup urine disease, type Ia, 248600 |
| *BCKDHB* | 99,50% | 94,40% | 100% | 100% | Maple syrup urine disease, type Ib, 248600 |
| *BCKDK* | 100% | 100% | 100% | 100% | Branched-chain ketoacid dehydrogenase kinase deficiency, 614923 |

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| *BCO1* | 100% | 100% | 100% | 100% | ?Hypercarotenemia and vitamin A deficiency, autosomal dominant, 115300 |
| *BLVRA* | 100% | 99,40% | 100% | 100% | Hyperbiliverdinemia, 614156 |
| *BMP2* | 100% | 100% | 100% | 100% | Brachydactyly, type A2, 112600<br>Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies, 617877 |
| *BPGM* | 100% | 100% | 100% | 100% | Erythrocytosis, familial, 8, 222800 |
| *BTD* | 100% | 99,90% | 100% | 100% | Biotinidase deficiency, 253260 |
| *C1GALT1C1* | 100% | 99,50% | 100% | 100% | Tn polyagglutination syndrome, somatic, 300622 |
| *CA5A* | 99,70% | 97,10% | 100% | 100% | Hyperammonemia due to carbonic anhydrase VA deficiency, 615751 |
| *CAD* | 100% | 99,20% | 100% | 100% | Epileptic encephalopathy, early infantile, 50, 616457 |
| *CANT1* | 100% | 99,90% | 100% | 100% | Desbuquois dysplasia 1, 251450<br>Epiphyseal dysplasia, multiple, 7, 617719 |
| *CAT* | 100% | 100% | 100% | 100% | Acatalasemia, 614097 |
| *CBS* | 99,80% | 98,30% | 100% | 100% | Homocystinuria, B6-responsive and nonresponsive types, 236200<br>Thrombosis, hyperhomocysteinemic, 236200 |
| *CCDC115* | 95,30% | 90,00% | 100% | 100% | Congenital disorder of glycosylation, type IIo, 616828 |
| *CEL* | 89,70% | 88,00% | 100% | 99,80% | Maturity-onset diabetes of the young, type VIII, 609812 |
| *CERKL* | 99,50% | 96,90% | 100% | 100% | Retinitis pigmentosa 26, 608380 |
| *CERS3* | 99,90% | 98,90% | 100% | 100% | Ichthyosis, congenital, autosomal recessive 9, 615023 |
| *CFTR* | 99,60% | 97,90% | 100% | 100% | Cystic fibrosis, 219700<br>Congenital bilateral absence of vas deferens, 277180<br>Sweat chloride elevation without CF, 0 |
| *CHIT1* | 99,70% | 98,10% | 100% | 100% | No OMIM disease ID |
| *CHKB* | 100% | 99,70% | 100% | 100% | Muscular dystrophy, congenital, megaconial type, 602541 |
| *CHST14* | 99,90% | 98,90% | 100% | 100% | Ehlers-Danlos syndrome, musculocontractural type 1, 601776 |
| *CHST3* | 100% | 99,40% | 100% | 100% | Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095 |
| *CHST6* | 100% | 100% | 100% | 100% | Macular corneal dystrophy, 217800 |
| *CHSY1* | 97,20% | 95,70% | 99,70% | 98,00% | Temtamy preaxial brachydactyly syndrome, 605282 |
| *CLCN7* | 99,70% | 98,40% | 100% | 100% | Osteopetrosis, autosomal recessive 4, 611490<br>Osteopetrosis, autosomal dominant 2, 166600<br>Hypopigmentation, organomegaly, and delayed myelination and development, 618541 |
| *CLN3* | 92,50% | 91,80% | 92,50% | 92,50% | Ceroid lipofuscinosis, neuronal, 3, 204200 |
| *CLN5* | 99,40% | 95,50% | 100% | 100% | Ceroid lipofuscinosis, neuronal, 5, 256731 |
| *CLN6* | 99,90% | 97,10% | 100% | 100% | Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300<br>Ceroid lipofuscinosis, neuronal, 6, 601780 |
| *CLN8* | 83,50% | 83,50% | 100% | 100% | Ceroid lipofuscinosis, neuronal, 8, 600143<br>Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003 |
| *CLPB* | 100% | 99,90% | 100% | 100% | 3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271 |

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| *CMAS* | 99,90% | 97,80% | 100% | 100% | No OMIM disease ID |
| *COG1* | 100% | 100% | 100% | 100% | Congenital disorder of glycosylation, type IIg, 611209 |
| *COG2* | 99,90% | 98,50% | 100% | 100% | ?Congenital disorder of glycosylation, type IIq, 617395 |
| *COG4* | 100% | 99,90% | 100% | 100% | Saul-Wilson syndrome, 618150<br>Congenital disorder of glycosylation, type IIj, 613489 |
| *COG5* | 99,70% | 97,60% | 100% | 100% | Congenital disorder of glycosylation, type IIi, 613612 |
| *COG6* | 99,10% | 93,90% | 100% | 100% | Shaheen syndrome, 615328<br>Congenital disorder of glycosylation, type III, 614576 |
| *COG7* | 100% | 100% | 100% | 100% | Congenital disorder of glycosylation, type IIe, 608779 |
| *COG8* | 100% | 99,60% | 100% | 100% | Congenital disorder of glycosylation, type IIh, 611182 |
| *COMT* | 100% | 99,90% | 100% | 100% | No OMIM disease ID |
| *COQ2* | 98,00% | 95,30% | 97,20% | 97,20% | Coenzyme Q10 deficiency, primary, 1, 607426 |
| *COQ4* | 90,90% | 89,30% | 100% | 100% | Coenzyme Q10 deficiency, primary, 7, 616276 |
| *COQ5* | 100% | 100% | 100% | 100% | No OMIM disease ID |
| *COQ6* | 99,90% | 98,40% | 100% | 100% | Coenzyme Q10 deficiency, primary, 6, 614650 |
| *COQ7* | 100% | 99,80% | 100% | 100% | ?Coenzyme Q10 deficiency, primary, 8, 616733 |
| *COQ8A* | 100% | 99,50% | 100% | 100% | Coenzyme Q10 deficiency, primary, 4, 612016 |
| *COQ8B* | 100% | 99,30% | 100% | 100% | Nephrotic syndrome, type 9, 615573 |
| *COQ9* | 100% | 97,90% | 100% | 100% | Coenzyme Q10 deficiency, primary, 5, 614654 |
| *CP* | 94,80% | 88,90% | 100% | 100% | Hemosiderosis, systemic, due to aceruloplasminemia, 604290<br>Cerebellar ataxia, 604290 |
| *CPOX* | 99,90% | 95,40% | 100% | 100% | Harderoporphyria, 121300<br>Coproporphyria, 121300 |
| *CPS1* | 100% | 99,90% | 100% | 100% | Carbamoylphosphate synthetase I deficiency, 237300 |
| *CPT1A* | 100% | 98,90% | 100% | 100% | CPT deficiency, hepatic, type IA, 255120 |
| *CPT2* | 98,20% | 97,80% | 100% | 100% | CPT II deficiency, myopathic, stress-induced, 255110<br>CPT II deficiency, infantile, 600649<br>CPT II deficiency, lethal neonatal, 608836 |
| *CRAT* | 100% | 99,80% | 100% | 100% | ?Neurodegeneration with brain iron accumulation 8, 617917 |
| *CTH* | 100% | 100% | 100% | 100% | Cystathioninuria, 219500<br>Homocysteine, total plasma, elevated, 0 |
| *CTNS* | 100% | 99,80% | 100% | 100% | Cystinosis, nephropathic, 219800<br>Cystinosis, ocular nonnephropathic, 219750<br>Cystinosis, late-onset juvenile or adolescent nephropathic, 219900<br>Cystinosis, atypical nephropathic, 219800 |
| *CTSA* | 100% | 100% | 100% | 100% | Galactosialidosis, 256540 |

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| *CTSC* | 100% | 100% | 100% | 100% | Periodontitis 1, juvenile, 170650<br>Papillon-Lefevre syndrome, 245000<br>Haim-Munk syndrome, 245010 |
| *CTSD* | 98,40% | 95,00% | 100% | 100% | Ceroid lipofuscinosis, neuronal, 10, 610127 |
| *CTSK* | 100% | 99,90% | 100% | 100% | Pycnodysostosis, 265800 |
| *CUBN* | 99,70% | 98,30% | 100% | 100% | Megaloblastic anemia-1, Finnish type, 261100 |
| *CYB561* | 92,80% | 92,60% | 100% | 99,90% | Orthostatic hypotension 2, 618182 |
| *CYB5R3* | 98,40% | 98,00% | 99,80% | 98,90% | Methemoglobinemia, type I, 250800<br>Methemoglobinemia, type II, 250800 |
| *CYP11A1* | 99,30% | 96,10% | 100% | 100% | Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743 |
| *CYP11B1* | 100% | 100% | 100% | 100% | Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010<br>Aldosteronism, glucocorticoid-remediable, 103900 |
| *CYP11B2* | 100% | 100% | 100% | 100% | Hypoaldosteronism, congenital, due to CMO II deficiency, 610600<br>Hypoaldosteronism, congenital, due to CMO I deficiency, 203400<br>Aldosterone to renin ratio raised, 0 |
| *CYP17A1* | 100% | 99,50% | 100% | 100% | 17-alpha-hydroxylase/17,20-lyase deficiency, 202110<br>17,20-lyase deficiency, isolated, 202110 |
| *CYP19A1* | 98,80% | 96,80% | 100% | 100% | Aromatase deficiency, 613546<br>Aromatase excess syndrome, 139300 |
| *CYP1B1* | 100% | 100% | 100% | 100% | Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300<br>Anterior segment dysgenesis 6, multiple subtypes, 617315 |
| *CYP21A2* | 97,80% | 88,40% | 100% | 100% | Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910<br>Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910 |
| *CYP27A1* | 98,90% | 96,70% | 100% | 100% | Cerebrotendinous xanthomatosis, 213700 |
| *CYP27B1* | 99,90% | 99,30% | 100% | 100% | Vitamin D-dependent rickets, type I, 264700 |
| *CYP2R1* | 99,40% | 95,60% | 100% | 100% | Rickets due to defect in vitamin D 25-hydroxylation, 600081 |
| *CYP2U1* | 94,80% | 91,50% | 100% | 99,90% | Spastic paraplegia 56, autosomal recessive, 615030 |
| *CYP7B1* | 98,00% | 92,80% | 100% | 100% | Spastic paraplegia 5A, autosomal recessive, 270800<br>Bile acid synthesis defect, congenital, 3, 613812 |
| *D2HGDH* | 99,20% | 97,20% | 100% | 100% | D-2-hydroxyglutaric aciduria, 600721 |
| *DAO* | 100% | 100% | 100% | 100% | No OMIM disease ID |
| *DBH* | 100% | 100% | 100% | 100% | Orthostatic hypotension 1, due to DBH deficiency, 223360 |
| *DBT* | 99,80% | 98,00% | 100% | 100% | Maple syrup urine disease, type II, 248600 |
| *DCXR* | 98,60% | 93,60% | 100% | 100% | No OMIM disease ID |
| *DDC* | 99,70% | 96,40% | 100% | 100% | Aromatic L-amino acid decarboxylase deficiency, 608643 |
| *DDHD1* | 97,90% | 95,80% | 100% | 100% | Spastic paraplegia 28, autosomal recessive, 609340 |
| *DDOST* | 100% | 99,90% | 100% | 100% | ?Congenital disorder of glycosylation, type Ir, 614507 |

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| *DEGS1* | 100% | 100% | 100% | 100% | Leukodystrophy, hypomyelinating, 18, 618404 |
| *DGAT1* | 91,90% | 87,60% | 99,70% | 98,60% | ?Diarrhea 7, protein-losing enteropathy type, 615863 |
| *DGKE* | 99,80% | 98,10% | 100% | 100% | Nephrotic syndrome, type 7, 615008 |
| *DGUOK* | 100% | 99,40% | 100% | 100% | Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, 617070<br>Portal hypertension, noncirrhotic, 617068<br>Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880 |
| *DHCR24* | 100% | 100% | 100% | 100% | Desmosterolosis, 602398 |
| *DHCR7* | 100% | 100% | 100% | 100% | Smith-Lemli-Opitz syndrome, 270400 |
| *DHDDS* | 99,00% | 95,00% | 95,20% | 95,20% | Retinitis pigmentosa 59, 613861<br>?Congenital disorder of glycosylation, type 1bb, 613861<br>Developmental delay and seizures with or without movement abnormalities, 617836 |
| *DHFR* | 92,10% | 78,90% | 100% | 100% | Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839 |
| *DHODH* | 100% | 100% | 100% | 100% | Miller syndrome, 263750 |
| *DLD* | 100% | 99,70% | 100% | 100% | Dihydrolipoamide dehydrogenase deficiency, 246900 |
| *DMGDH* | 100% | 99,70% | 100% | 100% | Dimethylglycine dehydrogenase deficiency, 605850 |
| *DNAJC12* | 87,40% | 87,40% | 100% | 100% | Hyperphenylalaninemia, mild, non-BH4-deficient, 617384 |
| *DNAJC19* | 98,90% | 96,20% | 100% | 100% | 3-methylglutaconic aciduria, type V, 610198 |
| *DNM1L* | 99,90% | 98,50% | 100% | 100% | Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388<br>Optic atrophy 5, 610708 |
| *DNM2* | 98,10% | 94,50% | 100% | 100% | Lethal congenital contracture syndrome 5, 615368<br>Charcot-Marie-Tooth disease, axonal type 2M, 606482<br>Centronuclear myopathy 1, 160150<br>Charcot-Marie-Tooth disease, dominant intermediate B, 606482 |
| *DNMT1* | 99,20% | 99,00% | 99,70% | 99,20% | Neuropathy, hereditary sensory, type IE, 614116<br>Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121 |
| *DNMT3B* | 100% | 100% | 100% | 100% | Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860 |
| *DOLK* | 100% | 100% | 100% | 100% | Congenital disorder of glycosylation, type Im, 610768 |
| *DPAGT1* | 100% | 100% | 100% | 100% | Congenital disorder of glycosylation, type Ij, 608093<br>Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750 |
| *DPM1* | 98,20% | 91,30% | 99,70% | 97,10% | Congenital disorder of glycosylation, type Ie, 608799 |
| *DPM2* | 100% | 98,70% | 100% | 100% | Congenital disorder of glycosylation, type Iu, 615042 |
| *DPM3* | 100% | 100% | 100% | 100% | Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 15, 612937 |
| *DPYD* | 99,70% | 97,70% | 100% | 100% | Dihydropyrimidine dehydrogenase deficiency, 274270<br>5-fluorouracil toxicity, 274270 |
| *DPYS* | 100% | 99,90% | 100% | 100% | Dihydropyrimidinuria, 222748 |
| *DTYMK* | 100% | 99,80% | 100% | 100% | No OMIM disease ID |
| *EBP* | 99,70% | 95,80% | 100% | 100% | Chondrodysplasia punctata, X-linked dominant, 302960<br>MEND syndrome, 300960 |

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| *ECHS1* | 99,90% | 99,00% | 100% | 100% | Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277 |
| *ELOVL1* | 99,80% | 97,60% | 100% | 100% | Ichthyotic keratoderma, spasticity, hypomyelination, and dysmorphic facies, 618527 |
| *ELOVL4* | 100% | 99,50% | 100% | 100% | Spinocerebellar ataxia 34, 133190<br>Stargardt disease 3, 600110<br>Ichthyosis, spastic quadriplegia, and mental retardation, 614457 |
| *ENO3* | 100% | 99,90% | 100% | 100% | ?Glycogen storage disease XIII, 612932 |
| *EOGT* | 79,40% | 78,40% | 91,90% | 89,00% | Adams-Oliver syndrome 4, 615297 |
| *EPHX1* | 99,90% | 98,80% | 100% | 100% | ?Hypercholanemia, familial, 607748 |
| *EPHX2* | 99,50% | 96,20% | 100% | 100% | No OMIM disease ID |
| *ETFA* | 100% | 100% | 100% | 100% | Glutaric acidemia IIA, 231680 |
| *ETFB* | 100% | 99,80% | 100% | 100% | Glutaric acidemia IIB, 231680 |
| *ETFDH* | 100% | 99,80% | 100% | 100% | Glutaric acidemia IIC, 231680 |
| *ETHE1* | 99,90% | 97,40% | 100% | 100% | Ethylmalonic encephalopathy, 602473 |
| *EXT1* | 99,90% | 98,40% | 100% | 100% | Exostoses, multiple, type 1, 133700<br>Chondrosarcoma, 215300 |
| *EXT2* | 100% | 99,30% | 100% | 100% | Exostoses, multiple, type 2, 133701<br>Seizures, scoliosis, and macrocephaly syndrome, 616682 |
| *FA2H* | 92,00% | 83,10% | 100% | 100% | Spastic paraplegia 35, autosomal recessive, 612319 |
| *FAH* | 100% | 100% | 100% | 100% | Tyrosinemia, type I, 276700 |
| *FAR1* | 97,60% | 92,80% | 100% | 100% | Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154 |
| *FBP1* | 100% | 99,50% | 100% | 100% | Fructose-1,6-bisphosphatase deficiency, 229700 |
| *FDFT1* | 97,70% | 96,00% | 100% | 100% | Squalene synthase deficiency, 618156 |
| *FECH* | 100% | 100% | 100% | 100% | Protoporphyria, erythropoietic, 1, 177000 |
| *FH* | 92,10% | 88,30% | 100% | 100% | Fumarase deficiency, 606812<br>Leiomyomatosis and renal cell cancer, 150800 |
| *FKRP* | 100% | 100% | 100% | 99,90% | Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155<br>Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, 606612<br>Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153 |
| *FKTN* | 99,70% | 97,00% | 100% | 100% | Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588<br>Cardiomyopathy, dilated, 1X, 611615<br>Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152<br>Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800 |
| *FLAD1* | 100% | 99,80% | 100% | 100% | Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency, 255100 |
| *FMO3* | 99,90% | 99,70% | 100% | 100% | Trimethylaminuria, 602079 |
| *FOLR1* | 100% | 100% | 100% | 100% | Neurodegeneration due to cerebral folate transport deficiency, 613068 |
| *FTCD* | 95,70% | 91,00% | 100% | 100% | Glutamate formiminotransferase deficiency, 229100 |

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| *FUCA1* | 100% | 99,90% | 100% | 100% | Fucosidosis, 230000 |
| *FUK* | 97,70% | 95,40% | 100% | 100% | Congenital disorder of glycosylation with defective fucosylation 2, 618324 |
| *FUT2* | 100% | 100% | 100% | 100% | No OMIM disease ID |
| *FUT6* | 100% | 100% | 100% | 100% | Fucosyltransferase 6 deficiency, 613852 |
| *FUT8* | 100% | 99,20% | 100% | 100% | Congenital disorder of glycosylation with defective fucosylation 1, 618005 |
| *G6PC* | 100% | 100% | 100% | 100% | Glycogen storage disease Ia, 232200 |
| *G6PC3* | 100% | 99,90% | 100% | 100% | Dursun syndrome, 612541<br>Neutropenia, severe congenital 4, autosomal recessive, 612541 |
| *G6PD* | 99,30% | 98,10% | 100% | 99,30% | Hemolytic anemia, G6PD deficient (favism), 300908 |
| *GAA* | 100% | 99,90% | 100% | 100% | Glycogen storage disease II, 232300 |
| *GAD1* | 100% | 99,90% | 100% | 100% | ?Cerebral palsy, spastic quadriplegic, 1, 603513 |
| *GALC* | 99,80% | 98,30% | 100% | 100% | Krabbe disease, 245200 |
| *GALE* | 100% | 100% | 100% | 100% | Galactose epimerase deficiency, 230350 |
| *GALK1* | 100% | 99,10% | 100% | 100% | Galactokinase deficiency with cataracts, 230200 |
| *GALM* | 100% | 99,90% | 100% | 100% | No OMIM disease ID |
| *GALNS* | 100% | 99,80% | 100% | 100% | Mucopolysaccharidosis IVA, 253000 |
| *GALNT3* | 99,80% | 99,00% | 100% | 100% | Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900 |
| *GALT* | 100% | 99,70% | 100% | 100% | Galactosemia, 230400 |
| *GAMT* | 93,10% | 82,70% | 100% | 100% | Cerebral creatine deficiency syndrome 2, 612736 |
| *GANAB* | 99,90% | 99,00% | 100% | 100% | Polycystic kidney disease 3, 600666 |
| *GATM* | 100% | 100% | 100% | 100% | Cerebral creatine deficiency syndrome 3, 612718 |
| *GBA* | 100% | 100% | 100% | 100% | Gaucher disease, type III, 231000<br>Gaucher disease, type IIIC, 231005<br>Gaucher disease, type I, 230800<br>Gaucher disease, perinatal lethal, 608013<br>Gaucher disease, type II, 230900 |
| *GBA2* | 100% | 99,70% | 100% | 100% | Spastic paraplegia 46, autosomal recessive, 614409 |
| *GBE1* | 100% | 99,60% | 100% | 100% | Polyglucosan body disease, adult form, 263570<br>Glycogen storage disease IV, 232500 |
| *GCDH* | 100% | 99,20% | 100% | 100% | Glutaricaciduria, type I, 231670 |
| *GCH1* | 99,90% | 95,50% | 100% | 100% | Hyperphenylalaninemia, BH4-deficient, B, 233910<br>Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 |
| *GCK* | 100% | 100% | 100% | 100% | Diabetes mellitus, noninsulin-dependent, late onset, 125853<br>Diabetes mellitus, permanent neonatal, 606176<br>MODY, type II, 125851<br>Hyperinsulinemic hypoglycemia, familial, 3, 602485 |
| *GCLC* | 99,80% | 98,00% | 100% | 100% | Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency, 230450 |

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| *GCLM* | 99,60% | 95,80% | 100% | 100% | No OMIM disease ID |
| *GCSH* | 75,70% | 68,90% | 100% | 100% | ?Glycine encephalopathy, 605899 |
| *GFPT1* | 100% | 99,40% | 100% | 100% | Myasthenia, congenital, 12, with tubular aggregates, 610542 |
| *GIF* | 100% | 99,70% | 100% | 100% | Intrinsic factor deficiency, 261000 |
| *GK* | 88,90% | 70,40% | 100% | 99,90% | Glycerol kinase deficiency, 307030 |
| *GLA* | 99,80% | 96,60% | 100% | 100% | Fabry disease, 301500<br>Fabry disease, cardiac variant, 301500 |
| *GLB1* | 99,90% | 97,40% | 100% | 100% | GM1-gangliosidosis, type III, 230650<br>GM1-gangliosidosis, type I, 230500<br>Mucopolysaccharidosis type IVB (Morquio), 253010<br>GM1-gangliosidosis, type II, 230600 |
| *GLDC* | 89,90% | 82,00% | 100% | 99,90% | Glycine encephalopathy, 605899 |
| *GLRA1* | 100% | 99,80% | 100% | 100% | Hyperekplexia 1, 149400 |
| *GLRX5* | 97,30% | 89,10% | 99,60% | 95,40% | Anemia, sideroblastic, 3, pyridoxine-refractory, 616860<br>Spasticity, childhood-onset, with hyperglycinemia, 616859 |
| *GLS* | 96,30% | 87,20% | 100% | 99,90% | ?Infantile cataract, skin abnormalities, glutamate excess, and impaired intellectual development, 618339<br>Global developmental delay, progressive ataxia, and elevated glutamine, 618412<br>Epileptic encephalopathy, early infantile, 71, 618328 |
| *GLUD1* | 94,20% | 82,90% | 100% | 100% | Hyperinsulinism-hyperammonemia syndrome, 606762 |
| *GLUL* | 99,90% | 98,20% | 100% | 100% | Glutamine deficiency, congenital, 610015 |
| *GLYCTK* | 98,80% | 97,30% | 100% | 100% | D-glyceric aciduria, 220120 |
| *GM2A* | 100% | 100% | 100% | 100% | GM2-gangliosidosis, AB variant, 272750 |
| *GMPPA* | 100% | 100% | 100% | 100% | Alacrima, achalasia, and mental retardation syndrome, 615510 |
| *GMPPB* | 100% | 100% | 100% | 100% | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350<br>Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14, 615351<br>Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352 |
| *GMPS* | 99,10% | 96,10% | 100% | 100% | No OMIM disease ID |
| *GNE* | 100% | 99,70% | 100% | 100% | Sialuria, 269921<br>Nonaka myopathy, 605820 |
| *GNMT* | 100% | 100% | 100% | 100% | Glycine N-methyltransferase deficiency, 606664 |
| *GNPAT* | 99,70% | 97,30% | 100% | 100% | Rhizomelic chondrodysplasia punctata, type 2, 222765 |
| *GNPTAB* | 100% | 99,90% | 100% | 100% | Mucolipidosis II alpha/beta, 252500<br>Mucolipidosis III alpha/beta, 252600 |
| *GNPTG* | 99,10% | 94,30% | 100% | 99,90% | Mucolipidosis III gamma, 252605 |
| *GNS* | 98,40% | 94,80% | 100% | 100% | Mucopolysaccharidosis type IIID, 252940 |
| *GOT1* | 100% | 99,30% | 100% | 100% | Aspartate aminotransferase, serum level of, QTL1, 614419 |
| *GOT2* | 97,50% | 90,90% | 100% | 100% | Epileptic encephalopathy, early infantile, 82, 618721 |

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| *GPD1* | 100% | 100% | 100% | 100% | Hypertriglyceridemia, transient infantile, 614480 |
| *GPD1L* | 100% | 99,80% | 100% | 100% | Brugada syndrome 2, 611777 |
| *GPHN* | 100% | 99,50% | 100% | 100% | Molybdenum cofactor deficiency C, 615501 |
| *GPI* | 100% | 100% | 100% | 100% | Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency, 613470 |
| *GPT2* | 99,20% | 93,60% | 100% | 99,80% | Mental retardation, autosomal recessive 49, 616281 |
| *GPX1* | 95,40% | 84,60% | 100% | 100% | No OMIM disease ID |
| *GRHPR* | 84,20% | 81,30% | 100% | 99,30% | Hyperoxaluria, primary, type II, 260000 |
| *GSS* | 100% | 99,90% | 100% | 100% | Glutathione synthetase deficiency, 266130<br>Hemolytic anemia due to glutathione synthetase deficiency, 231900 |
| *GUSB* | 92,90% | 91,70% | 100% | 100% | Mucopolysaccharidosis VII, 253220 |
| *GYG1* | 99,90% | 99,20% | 100% | 100% | ?Glycogen storage disease XV, 613507<br>Polyglucosan body myopathy 2, 616199 |
| *GYS1* | 100% | 98,60% | 100% | 100% | Glycogen storage disease 0, muscle, 611556 |
| *GYS2* | 99,80% | 99,00% | 100% | 100% | Glycogen storage disease 0, liver, 240600 |
| *H6PD* | 99,00% | 99,00% | 100% | 100% | Cortisone reductase deficiency 1, 604931 |
| *HADH* | 99,00% | 97,50% | 100% | 100% | 3-hydroxyacyl-CoA dehydrogenase deficiency, 231530<br>Hyperinsulinemic hypoglycemia, familial, 4, 609975 |
| *HADHA* | 97,10% | 91,30% | 100% | 100% | LCHAD deficiency, 609016<br>HELLP syndrome, maternal, of pregnancy, 609016<br>Fatty liver, acute, of pregnancy, 609016<br>Trifunctional protein deficiency, 609015 |
| *HADHB* | 98,80% | 89,70% | 100% | 100% | Trifunctional protein deficiency, 609015 |
| *HAGH* | 100% | 99,70% | 98,70% | 96,10% | No OMIM disease ID |
| *HEXA* | 93,80% | 93,30% | 100% | 100% | GM2-gangliosidosis, several forms, 272800<br>Tay-Sachs disease, 272800 |
| *HEXB* | 99,60% | 96,90% | 100% | 99,90% | Sandhoff disease, infantile, juvenile, and adult forms, 268800 |
| *HFE* | 100% | 99,70% | 100% | 100% | Hemochromatosis, 235200 |
| *HGD* | 100% | 100% | 100% | 100% | Alkaptonuria, 203500 |
| *HGSNAT* | 86,40% | 86,30% | 91,20% | 89,30% | Retinitis pigmentosa 73, 616544<br>Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 |
| *HIBADH* | 94,40% | 91,20% | 100% | 100% | No OMIM disease ID |
| *HIBCH* | 98,20% | 88,50% | 100% | 100% | 3-hydroxyisobutryl-CoA hydrolase deficiency, 250620 |
| *HK1* | 100% | 100% | 100% | 100% | Hemolytic anemia due to hexokinase deficiency, 235700<br>Neuropathy, hereditary motor and sensory, Russe type, 605285<br>Neurodevelopmental disorder with visual defects and brain anomalies, 618547<br>Retinitis pigmentosa 79, 617460 |
| *HLCS* | 100% | 100% | 100% | 100% | Holocarboxylase synthetase deficiency, 253270 |

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| *HMBS* | 99,90% | 99,40% | 100% | 100% | Porphyria, acute intermittent, 176000<br>Porphyria, acute intermittent, nonerythroid variant, 176000 |
| *HMGCL* | 100% | 99,80% | 100% | 100% | HMG-CoA lyase deficiency, 246450 |
| *HMGCS2* | 100% | 99,60% | 100% | 100% | HMG-CoA synthase-2 deficiency, 605911 |
| *HMOX1* | 98,40% | 89,90% | 100% | 100% | Heme oxygenase-1 deficiency, 614034 |
| *HNF1A* | 100% | 99,80% | 100% | 100% | MODY, type III, 600496<br>Hepatic adenoma, somatic, 142330<br>Renal cell carcinoma, 144700<br>Diabetes mellitus, insulin-dependent, 20, 612520 |
| *HNF4A* | 99,90% | 99,00% | 100% | 100% | MODY, type I, 125850<br>Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young, 616026 |
| *HOGA1* | 100% | 96,40% | 100% | 100% | Hyperoxaluria, primary, type III, 613616 |
| *HPD* | 100% | 100% | 100% | 100% | Tyrosinemia, type III, 276710<br>Hawkinsinuria, 140350 |
| *HPRT1* | 99,30% | 91,80% | 100% | 99,30% | HPRT-related gout, 300323<br>Lesch-Nyhan syndrome, 300322 |
| *HS6ST1* | 92,90% | 84,50% | 100% | 100% | No OMIM disease ID |
| *HSD11B1* | 100% | 99,60% | 100% | 100% | Cortisone reductase deficiency 2, 614662 |
| *HSD11B2* | 86,00% | 82,70% | 99,90% | 98,10% | Apparent mineralocorticoid excess, 218030 |
| *HSD17B10* | 100% | 99,10% | 100% | 100% | HSD10 mitochondrial disease, 300438 |
| *HSD17B3* | 100% | 100% | 100% | 100% | Pseudohermaphroditism, male, with gynecomastia, 264300 |
| *HSD17B4* | 96,00% | 93,70% | 96,60% | 96,60% | D-bifunctional protein deficiency, 261515<br>Perrault syndrome 1, 233400 |
| *HSD3B2* | 100% | 99,70% | 100% | 100% | Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency, 201810 |
| *HSD3B7* | 99,10% | 95,50% | 100% | 100% | Bile acid synthesis defect, congenital, 1, 607765 |
| *HTRA2* | 100% | 99,90% | 100% | 100% | 3-methylglutaconic aciduria, type VIII, 617248 |
| *HYAL1* | 100% | 100% | 100% | 100% | ?Mucopolysaccharidosis type IX, 601492 |
| *IDH2* | 99,70% | 97,40% | 100% | 99,80% | D-2-hydroxyglutaric aciduria 2, 613657 |
| *IDH3B* | 95,40% | 95,40% | 100% | 100% | Retinitis pigmentosa 46, 612572 |
| *IDI1* | 99,80% | 97,60% | 100% | 100% | No OMIM disease ID |
| *IDS* | 99,90% | 98,00% | 100% | 100% | Mucopolysaccharidosis II, 309900 |
| *IDUA* | 93,70% | 86,80% | 100% | 100% | Mucopolysaccharidosis Ih/s, 607015<br>Mucopolysaccharidosis Ih, 607014<br>Mucopolysaccharidosis Is, 607016 |
| *IMPAD1* | 100% | 100% | 100% | 100% | Chondrodysplasia with joint dislocations, GPAPP type, 614078 |
| *IMPDH1* | 87,90% | 80,20% | 100% | 100% | Leber congenital amaurosis 11, 613837<br>Retinitis pigmentosa 10, 180105 |

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| *INPP5E* | 97,10% | 92,70% | 100% | 100% | Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156<br>Joubert syndrome 1, 213300 |
| *INPPL1* | 98,40% | 94,50% | 99,90% | 99,70% | Opsismodysplasia, 258480 |
| *INSR* | 97,80% | 94,70% | 99,90% | 99,20% | Hyperinsulinemic hypoglycemia, familial, 5, 609968<br>Rabson-Mendenhall syndrome, 262190<br>Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549<br>Leprechaunism, 246200 |
| *MR PL44* | 99,90% | 98,70% | 100% | 100% | ?Combined oxidative phosphorylation deficiency 16, 615395 |
| *MR PS36* | 95,20% | 77,60% | 100% | 100% | No OMIM disease ID |
| *IREB2* | 100% | 99,80% | 100% | 100% | Neurodegeneration, early-onset, with choreoathetoid movements and microcytic anemia, 618451 |
| *ISPD* | 98,50% | 94,80% | 100% | 99,40% | Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052<br>Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643 |
| *ITCH* | 95,40% | 95,20% | 100% | 99,00% | Autoimmune disease, multisystem, with facial dysmorphism, 613385 |
| *ITPA* | 100% | 100% | 100% | 100% | Epileptic encephalopathy, early infantile, 35, 616647 |
| *IVD* | 100% | 100% | 100% | 100% | Isovaleric acidemia, 243500 |
| *KCNA2* | 100% | 99,60% | 100% | 100% | Epileptic encephalopathy, early infantile, 32, 616366 |
| *KCNJ11* | 100% | 100% | 100% | 100% | Maturity-onset diabetes of the young, type 13, 616329<br>Diabetes, permanent neonatal, with or without neurologic features, 606176<br>Diabetes mellitus, transient neonatal, 3, 610582<br>Hyperinsulinemic hypoglycemia, familial, 2, 601820 |
| *KMT2A* | 100% | 99,90% | 99,90% | 99,40% | Wiedemann-Steiner syndrome, 605130 |
| *KMT2D* | 100% | 99,40% | 100% | 100% | Kabuki syndrome 1, 147920 |
| *L2HGDH* | 99,00% | 97,20% | 100% | 100% | L-2-hydroxyglutaric aciduria, 236792 |
| *LAMP2* | 99,20% | 95,60% | 100% | 100% | Danon disease, 300257 |
| *LARGE1* | 100% | 99,60% | 100% | 100% | Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6, 608840<br>Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154 |
| *LCAT* | 99,00% | 93,80% | 100% | 100% | Norum disease, 245900<br>Fish-eye disease, 136120 |
| *LCT* | 99,80% | 98,50% | 100% | 100% | Lactase deficiency, congenital, 223000 |
| *LDHA* | 95,00% | 91,70% | 100% | 100% | Glycogen storage disease XI, 612933 |
| *LDHB* | 96,30% | 86,00% | 100% | 100% | No OMIM disease ID |
| *LFNG* | 87,90% | 86,40% | 92,20% | 87,70% | Spondylocostal dysostosis 3, autosomal recessive, 609813 |
| *LIAS* | 100% | 99,10% | 100% | 100% | Hyperglycinemia, lactic acidosis, and seizures, 614462 |
| *LIPA* | 99,20% | 95,20% | 95,20% | 95,20% | Wolman disease, 278000<br>Cholesteryl ester storage disease, 278000 |
| *LIPC* | 100% | 99,40% | 100% | 100% | Hepatic lipase deficiency, 614025 |

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|---|---|---|---|---|---|
| *LIPT1* | 100% | 99,90% | 100% | 100% | Lipoyltransferase 1 deficiency, 616299 |
| *LIPT2* | 94,90% | 75,20% | 100% | 100% | Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities, 617668 |
| *LMBRD1* | 98,50% | 93,90% | 100% | 100% | Methylmalonic aciduria and homocystinuria, cblF type, 277380 |
| *LPIN1* | 99,60% | 97,30% | 100% | 100% | Myoglobinuria, acute recurrent, autosomal recessive, 268200 |
| *LPIN2* | 100% | 100% | 100% | 100% | Majeed syndrome, 609628 |
| *LPL* | 100% | 100% | 100% | 100% | Lipoprotein lipase deficiency, 238600<br>Combined hyperlipidemia, familial, 144250 |
| *LRAT* | 100% | 100% | 100% | 100% | Retinal dystrophy, early-onset severe, 613341<br>Leber congenital amaurosis 14, 613341<br>Retinitis pigmentosa, juvenile, 613341 |
| *LTC4S* | 74,20% | 68,50% | 100% | 100% | No OMIM disease ID |
| *LYST* | 99,60% | 98,30% | 100% | 100% | Chediak-Higashi syndrome, 214500 |
| *MAN1B1* | 100% | 99,70% | 100% | 99,90% | Mental retardation, autosomal recessive 15, 614202 |
| *MAN2B1* | 99,80% | 97,90% | 100% | 100% | Mannosidosis, alpha-, types I and II, 248500 |
| *MANBA* | 99,80% | 98,40% | 100% | 100% | Mannosidosis, beta, 248510 |
| *MAOA* | 100% | 99,70% | 99,80% | 98,50% | Brunner syndrome, 300615 |
| *MAT1A* | 99,70% | 97,70% | 100% | 100% | Methionine adenosyltransferase deficiency, autosomal recessive, 250850<br>Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850 |
| *MBOAT7* | 100% | 99,50% | 100% | 100% | Mental retardation, autosomal recessive 57, 617188 |
| *MCCC1* | 100% | 99,80% | 100% | 100% | 3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200 |
| *MCCC2* | 99,90% | 98,40% | 100% | 100% | 3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210 |
| *MCEE* | 100% | 100% | 100% | 100% | Methylmalonyl-CoA epimerase deficiency, 251120 |
| *MCOLN1* | 99,80% | 98,40% | 100% | 100% | Mucolipidosis IV, 252650 |
| *MDH1* | 100% | 99,40% | 100% | 100% | No OMIM disease ID |
| *MFSD2A* | 99,70% | 98,50% | 100% | 100% | Microcephaly 15, primary, autosomal recessive, 616486 |
| *MFSD8* | 100% | 99,70% | 100% | 100% | Macular dystrophy with central cone involvement, 616170<br>Ceroid lipofuscinosis, neuronal, 7, 610951 |
| *MGAT2* | 100% | 100% | 100% | 100% | Congenital disorder of glycosylation, type IIa, 212066 |
| *MINPP1* | 100% | 99,50% | 100% | 100% | No OMIM disease ID |
| *MLYCD* | 96,00% | 90,40% | 100% | 98,90% | Malonyl-CoA decarboxylase deficiency, 248360 |
| *MMAA* | 100% | 100% | 100% | 100% | Methylmalonic aciduria, vitamin B12-responsive, 251100 |
| *MMAB* | 100% | 99,60% | 100% | 100% | Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type, 251110 |
| *MMACHC* | 100% | 100% | 100% | 100% | Methylmalonic aciduria and homocystinuria, cblC type, 277400 |

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|---|---|---|---|---|---|
| *MMADHC* | 94,40% | 83,50% | 89,70% | 89,70% | Homocystinuria, cblD type, variant 1, 277410<br>Methylmalonic aciduria and homocystinuria, cblD type, 277410<br>Methylmalonic aciduria, cblD type, variant 2, 277410 |
| *MOCOS* | 99,80% | 97,70% | 100% | 100% | Xanthinuria, type II, 603592 |
| *MOCS1* | 99,20% | 95,40% | 100% | 100% | Molybdenum cofactor deficiency A, 252150 |
| *MOCS2* | 99,60% | 99,50% | 100% | 100% | Molybdenum cofactor deficiency B, 252160 |
| *MOGS* | 100% | 99,90% | 100% | 100% | Congenital disorder of glycosylation, type IIb, 606056 |
| *MPDU1* | 100% | 100% | 100% | 100% | Congenital disorder of glycosylation, type If, 609180 |
| *MPI* | 100% | 99,90% | 100% | 100% | Congenital disorder of glycosylation, type Ib, 602579 |
| *MSMO1* | 96,30% | 88,90% | 100% | 100% | Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834 |
| *MTHFD1* | 100% | 99,50% | 100% | 100% | Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia, 617780 |
| *MTHFR* | 97,30% | 96,00% | 100% | 100% | Homocystinuria due to MTHFR deficiency, 236250 |
| *MTMR 2* | 100% | 99,00% | 100% | 100% | Charcot-Marie-Tooth disease, type 4B1, 601382 |
| *MTM1* | 99,00% | 93,30% | 100% | 100% | Myotubular myopathy, X-linked, 310400 |
| *MTR* | 100% | 100% | 100% | 100% | Homocystinuria-megaloblastic anemia, cblG complementation type, 250940 |
| *MTRR* | 100% | 99,60% | 100% | 100% | Homocystinuria-megaloblastic anemia, cbl E type, 236270 |
| *MUT* | 99,80% | 98,30% | 100% | 100% | Methylmalonic aciduria, mut(0) type, 251000 |
| *MVK* | 90,90% | 90,50% | 90,50% | 90,50% | Hyper-IgD syndrome, 260920<br>Porokeratosis 3, multiple types, 175900<br>Mevalonic aciduria, 610377 |
| *NADK2* | 99,90% | 97,20% | 99,00% | 96,30% | 2,4-dienoyl-CoA reductase deficiency, 616034 |
| *NAGA* | 100% | 100% | 100% | 100% | Kanzaki disease, 609242<br>Schindler disease, type I, 609241<br>Schindler disease, type III, 609241 |
| *NAGLU* | 92,90% | 89,90% | 99,90% | 99,20% | Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920<br>?Charcot-Marie-Tooth disease, axonal, type 2V, 616491 |
| *NAGS* | 99,70% | 95,00% | 100% | 100% | N-acetylglutamate synthase deficiency, 237310 |
| *NANS* | 100% | 99,90% | 100% | 100% | Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442 |
| *NAXE* | 100% | 99,80% | 100% | 100% | Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186 |
| *NBAS* | 100% | 99,60% | 100% | 100% | Infantile liver failure syndrome 2, 616483<br>Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800 |
| *NEU1* | 99,70% | 97,70% | 100% | 100% | Sialidosis, type II, 256550<br>Sialidosis, type I, 256550 |
| *NGLY1* | 100% | 99,80% | 100% | 100% | Congenital disorder of deglycosylation, 615273 |
| *NMNAT1* | 100% | 99,20% | 98,30% | 95,60% | Leber congenital amaurosis 9, 608553 |
| *NNT* | 100% | 99,40% | 100% | 100% | Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency, 614736 |

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| *NPC1* | 99,60% | 98,70% | 100% | 100% | Niemann-Pick disease, type D, 257220<br>Niemann-Pick disease, type C1, 257220 |
| *NPC2* | 100% | 99,60% | 100% | 100% | Niemann-pick disease, type C2, 607625 |
| *NPL* | 100% | 99,90% | 100% | 100% | No OMIM disease ID |
| *NSD1* | 100% | 99,90% | 100% | 100% | Sotos syndrome 1, 117550 |
| *NSDHL* | 100% | 98,70% | 100% | 100% | CHILD syndrome, 308050<br>CK syndrome, 300831 |
| *NT5C3A* | 97,90% | 88,30% | 100% | 100% | Anemia, hemolytic, due to UMPH1 deficiency, 266120 |
| *NT5E* | 100% | 99,90% | 100% | 100% | Calcification of joints and arteries, 211800 |
| *NUS1* | 60,00% | 44,50% | 100% | 100% | Mental retardation, autosomal dominant 55, with seizures, 617831<br>?Congenital disorder of glycosylation, type 1aa, 617082 |
| *OAT* | 85,20% | 76,30% | 100% | 100% | Gyrate atrophy of choroid and retina with or without ornithinemia, 258870 |
| *OCRL* | 99,90% | 98,60% | 100% | 99,90% | Lowe syndrome, 309000<br>Dent disease 2, 300555 |
| *OPA3* | 100% | 99,00% | 100% | 100% | Optic atrophy 3 with cataract, 165300<br>3-methylglutaconic aciduria, type III, 258501 |
| *OPLAH* | 100% | 99,80% | 100% | 100% | 5-oxoprolinase deficiency, 260005 |
| *OTC* | 100% | 100% | 100% | 100% | Ornithine transcarbamylase deficiency, 311250 |
| *OXCT1* | 99,80% | 98,10% | 100% | 100% | Succinyl CoA:3-oxoacid CoA transferase deficiency, 245050 |
| *PAH* | 100% | 100% | 100% | 100% | Phenylketonuria, 261600 |
| *PANK2* | 100% | 99,30% | 100% | 100% | HARP syndrome, 607236<br>Neurodegeneration with brain iron accumulation 1, 234200 |
| *PC* | 99,80% | 97,30% | 100% | 100% | Pyruvate carboxylase deficiency, 266150 |
| *PCBD1* | 100% | 99,60% | 100% | 99,70% | Hyperphenylalaninemia, BH4-deficient, D, 264070 |
| *PCCA* | 99,50% | 96,70% | 100% | 100% | Propionicacidemia, 606054 |
| *PCCB* | 97,90% | 96,00% | 98,70% | 96,20% | Propionicacidemia, 606054 |
| *PCK1* | 100% | 100% | 100% | 100% | ?Phosphoenolpyruvate carboxykinase deficiency, cytosolic, 261680 |
| *PCK2* | 100% | 100% | 100% | 100% | No OMIM disease ID |
| *PCYT1A* | 98,90% | 95,50% | 100% | 100% | Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940 |
| *PCYT2* | 99,80% | 97,10% | 100% | 98,80% | Spastic paraplegia 82, autosomal recessive, 618770 |
| *PDSS1* | 94,70% | 87,60% | 97,30% | 96,60% | Coenzyme Q10 deficiency, primary, 2, 614651 |
| *PDSS2* | 99,80% | 97,10% | 100% | 100% | Coenzyme Q10 deficiency, primary, 3, 614652 |
| *PEPD* | 100% | 98,80% | 100% | 100% | Prolidase deficiency, 170100 |
| *PEX1* | 99,90% | 99,40% | 100% | 100% | Heimler syndrome 1, 234580<br>Peroxisome biogenesis disorder 1B (NALD/IRD), 601539<br>Peroxisome biogenesis disorder 1A (Zellweger), 214100 |

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| *PEX10* | 96,80% | 89,70% | 100% | 99,90% | Peroxisome biogenesis disorder 6B, 614871<br>Peroxisome biogenesis disorder 6A (Zellweger), 614870 |
| *PEX11B* | 100% | 99,60% | 100% | 100% | ?Peroxisome biogenesis disorder 14B, 614920 |
| *PEX12* | 100% | 100% | 100% | 100% | Peroxisome biogenesis disorder 3A (Zellweger), 614859<br>Peroxisome biogenesis disorder 3B, 266510 |
| *PEX13* | 100% | 100% | 100% | 100% | Peroxisome biogenesis disorder 11A (Zellweger), 614883<br>Peroxisome biogenesis disorder 11B, 614885 |
| *PEX14* | 96,70% | 90,80% | 100% | 100% | Peroxisome biogenesis disorder 13A (Zellweger), 614887 |
| *PEX16* | 97,90% | 94,20% | 100% | 100% | Peroxisome biogenesis disorder 8A (Zellweger), 614876<br>Peroxisome biogenesis disorder 8B, 614877 |
| *PEX19* | 99,90% | 98,50% | 100% | 100% | Peroxisome biogenesis disorder 12A (Zellweger), 614886 |
| *PEX2* | 100% | 100% | 100% | 100% | Peroxisome biogenesis disorder 5A (Zellweger), 614866<br>Peroxisome biogenesis disorder 5B, 614867 |
| *PEX26* | 100% | 100% | 100% | 100% | Peroxisome biogenesis disorder 7A (Zellweger), 614872<br>Peroxisome biogenesis disorder 7B, 614873 |
| *PEX3* | 100% | 99,30% | 100% | 100% | Peroxisome biogenesis disorder 10A (Zellweger), 614882<br>?Peroxisome biogenesis disorder 10B, 617370 |
| *PEX5* | 99,90% | 99,00% | 100% | 100% | Peroxisome biogenesis disorder 2B, 202370<br>Rhizomelic chondrodysplasia punctata, type 5, 616716<br>Peroxisome biogenesis disorder 2A (Zellweger), 214110 |
| *PEX6* | 94,50% | 86,70% | 100% | 100% | Peroxisome biogenesis disorder 4B, 614863<br>Heimler syndrome 2, 616617<br>Peroxisome biogenesis disorder 4A (Zellweger), 614862 |
| *PEX7* | 87,80% | 80,70% | 91,30% | 91,30% | Peroxisome biogenesis disorder 9B, 614879<br>Rhizomelic chondrodysplasia punctata, type 1, 215100 |
| *PFKM* | 100% | 99,50% | 100% | 100% | Glycogen storage disease VII, 232800 |
| *PGAM2* | 100% | 100% | 100% | 100% | Glycogen storage disease X, 261670 |
| *PGAP1* | 99,00% | 94,40% | 100% | 100% | Mental retardation, autosomal recessive 42, 615802 |
| *PGAP2* | 100% | 99,90% | 100% | 100% | Hyperphosphatasia with mental retardation syndrome 3, 614207 |
| *PGAP3* | 63,50% | 59,60% | 100% | 100% | Hyperphosphatasia with mental retardation syndrome 4, 615716 |
| *PGK1* | 92,80% | 79,30% | 100% | 100% | Phosphoglycerate kinase 1 deficiency, 300653 |
| *PGM1* | 100% | 100% | 100% | 100% | Congenital disorder of glycosylation, type It, 614921 |
| *PGM3* | 100% | 99,80% | 100% | 100% | Immunodeficiency 23, 615816 |
| *PHGDH* | 99,90% | 98,80% | 100% | 100% | Neu-Laxova syndrome 1, 256520<br>Phosphoglycerate dehydrogenase deficiency, 601815 |
| *PHKA1* | 99,20% | 95,30% | 100% | 99,90% | Muscle glycogenosis, 300559 |
| *PHKA2* | 100% | 99,70% | 100% | 99,60% | Glycogen storage disease, type IXa2, 306000<br>Glycogen storage disease, type IXa1, 306000 |

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| *PHKB* | 99,90% | 99,20% | 100% | 100% | Phosphorylase kinase deficiency of liver and muscle, autosomal recessive, 261750 |
| *PHKG1* | 99,90% | 97,80% | 100% | 100% | No OMIM disease ID |
| *PHKG2* | 100% | 99,90% | 100% | 100% | Glycogen storage disease IXc, 613027<br>Cirrhosis due to liver phosphorylase kinase deficiency, 0 |
| *PHYH* | 100% | 99,60% | 100% | 100% | Refsum disease, 266500 |
| *TALDO1* | 100% | 97,90% | 100% | 100% | Transaldolase deficiency, 606003 |
| *TANGO2* | 100% | 99,30% | 100% | 100% | metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878 |
| *TAT* | 100% | 100% | 100% | 100% | Tyrosinemia, type II, 276600 |
| *TAZ* | 99,20% | 96,50% | 100% | 100% | Barth syndrome, 302060 |
| *PIGA* | 93,80% | 86,70% | 100% | 100% | Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868<br>Paroxysmal nocturnal hemoglobinuria, somatic, 300818 |
| *PIGB* | 99,90% | 97,80% | 100% | 100% | Epileptic encephalopathy, early infantile, 80, 618580 |
| *PIGC* | 99,20% | 90,90% | 100% | 100% | Glycosylphosphatidylinositol biosynthesis defect 16, 617816 |
| *PIGL* | 100% | 100% | 100% | 100% | CHIME syndrome, 280000 |
| *PIGM* | 100% | 100% | 100% | 100% | Glycosylphosphatidylinositol deficiency, 610293 |
| *PIGN* | 93,80% | 91,50% | 98,80% | 98,80% | Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080 |
| *PIGO* | 100% | 99,90% | 100% | 100% | Hyperphosphatasia with mental retardation syndrome 2, 614749 |
| *PIGP* | 95,80% | 87,30% | 100% | 100% | Epileptic encephalopathy, early infantile, 55, 617599 |
| *PIGQ* | 92,80% | 90,80% | 100% | 100% | Epileptic encephalopathy, early infantile, 77, 618548 |
| *PIGT* | 98,10% | 98,10% | 100% | 100% | ?Paroxysmal nocturnal hemoglobinuria 2, 615399<br>Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398 |
| *PIGV* | 100% | 100% | 100% | 100% | Hyperphosphatasia with mental retardation syndrome 1, 239300 |
| *PIGW* | 100% | 99,80% | 100% | 100% | Glycosylphosphatidylinositol biosynthesis defect 11, 616025 |
| *PIGY* | 100% | 99,90% | 100% | 100% | Hyperphosphatasia with mental retardation syndrome 6, 616809 |
| *PIK3CA* | 100% | 99,80% | 100% | 100% | Ovarian cancer, somatic, 167000<br>Colorectal cancer, somatic, 114500<br>CLAPO syndrome, somatic, 613089<br>Cowden syndrome 5, 615108<br>Hepatocellular carcinoma, somatic, 114550<br>Breast cancer, somatic, 114480<br>Macrodactyly, somatic, 155500<br>Keratosis, seborrheic, somatic, 182000<br>Gastric cancer, somatic, 613659<br>Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501<br>Nevus, epidermal, somatic, 162900<br>CLOVE syndrome, somatic, 612918<br>Nonsmall cell lung cancer, somatic, 211980 |

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| *PIK3R1* | 99,80% | 99,00% | 100% | 100% | SHORT syndrome, 269880<br>Immunodeficiency 36, 616005<br>?Agammaglobulinemia 7, autosomal recessive, 615214 |
| *PIK3R2* | 90,70% | 89,60% | 99,30% | 96,10% | Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387 |
| *PIK3R5* | 100% | 99,90% | 100% | 100% | Ataxia-oculomotor apraxia 3, 615217 |
| *PIKFYVE* | 99,90% | 99,40% | 100% | 100% | Corneal fleck dystrophy, 121850 |
| *PIP5K1C* | 98,00% | 95,80% | 99,90% | 99,80% | Lethal congenital contractural syndrome 3, 611369 |
| *PKLR* | 100% | 99,20% | 100% | 100% | Pyruvate kinase deficiency, 266200<br>Adenosine triphosphate, elevated, of erythrocytes, 102900 |
| *PLA2G5* | 100% | 100% | 100% | 100% | No OMIM disease ID |
| *PLA2G6* | 99,90% | 98,30% | 100% | 100% | Infantile neuroaxonal dystrophy 1, 256600<br>Parkinson disease 14, autosomal recessive, 612953<br>Neurodegeneration with brain iron accumulation 2B, 610217 |
| *PLA2G7* | 99,90% | 99,00% | 100% | 100% | Platelet-activating factor acetylhydrolase deficiency, 614278 |
| *PLCB1* | 100% | 99,80% | 100% | 100% | Epileptic encephalopathy, early infantile, 12, 613722 |
| *PLCB4* | 99,90% | 98,80% | 100% | 100% | Auriculocondylar syndrome 2, 614669 |
| *PLCD1* | 99,90% | 97,80% | 100% | 100% | Nail disorder, nonsyndromic congenital, 3, (leukonychia), 151600 |
| *PLCE1* | 99,90% | 99,30% | 100% | 100% | Nephrotic syndrome, type 3, 610725 |
| *PLCG2* | 100% | 99,80% | 100% | 100% | Familial cold autoinflammatory syndrome 3, 614468<br>Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878 |
| *PLIN1* | 99,60% | 94,90% | 100% | 99,50% | Lipodystrophy, familial partial, type 4, 613877 |
| *PLOD1* | 100% | 98,40% | 100% | 100% | Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400 |
| *PLOD2* | 99,30% | 97,30% | 100% | 100% | Bruck syndrome 2, 609220 |
| *PLOD3* | 99,80% | 98,00% | 100% | 100% | Lysyl hydroxylase 3 deficiency, 612394 |
| *PLPBP* | 98,20% | 90,10% | 100% | 100% | Epilepsy, early-onset, vitamin B6-dependent, 617290 |
| *PMM2* | 100% | 100% | 100% | 100% | Congenital disorder of glycosylation, type Ia, 212065 |
| *PNLIP* | 100% | 99,80% | 100% | 100% | ?Pancreatic lipase deficiency, 614338 |
| *PNMT* | 99,60% | 96,70% | 100% | 100% | No OMIM disease ID |
| *PNP* | 99,80% | 98,90% | 100% | 100% | Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179 |
| *PNPLA2* | 99,70% | 96,10% | 100% | 100% | Neutral lipid storage disease with myopathy, 610717 |
| *PNPLA6* | 100% | 99,70% | 100% | 100% | Spastic paraplegia 39, autosomal recessive, 612020<br>Boucher-Neuhauser syndrome, 215470<br>Oliver-McFarlane syndrome, 275400<br>?Laurence-Moon syndrome, 245800 |
| *PNPO* | 99,90% | 97,70% | 100% | 100% | Pyridoxamine 5'-phosphate oxidase deficiency, 610090 |
| *POFUT1* | 100% | 99,00% | 100% | 100% | Dowling-Degos disease 2, 615327 |

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| *POGLUT1* | 99,40% | 94,60% | 100% | 100% | ?Muscular dystrophy, limb-girdle, autosomal recessive 21, 617232<br>Dowling-Degos disease 4, 615696 |
| *POLR3A* | 100% | 99,70% | 100% | 100% | Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694<br>Wiedemann-Rautenstrauch syndrome, 264090 |
| *POLR3B* | 99,90% | 98,60% | 100% | 100% | Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381 |
| *POMGNT1* | 100% | 99,90% | 100% | 100% | Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, 613151<br>Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157<br>Retinitis pigmentosa 76, 617123<br>Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280 |
| *POMGNT2* | 100% | 100% | 100% | 100% | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8, 614830<br>Muscular dystrophy-dystroglycanopathy (limb-girdle) type C, 8, 618135 |
| *POMK* | 100% | 100% | 100% | 100% | ?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094<br>Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249 |
| *POMT1* | 99,30% | 97,50% | 100% | 100% | Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308<br>Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670<br>Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155 |
| *POMT2* | 99,40% | 96,40% | 100% | 100% | Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158<br>Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150<br>Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2, 613156 |
| *PPCS* | 99,80% | 99,50% | 100% | 100% | Cardiomyopathy, dilated, 2C, 618189 |
| *PPM1K* | 100% | 100% | 100% | 100% | ?Maple syrup urine disease, mild variant, 615135 |
| *PPOX* | 99,70% | 96,80% | 100% | 100% | Porphyria variegata, 176200 |
| *PPT1* | 90,30% | 90,30% | 82,50% | 82,50% | Ceroid lipofuscinosis, neuronal, 1, 256730 |
| *PRKAG2* | 99,10% | 97,50% | 100% | 100% | Glycogen storage disease of heart, lethal congenital, 261740<br>Wolff-Parkinson-White syndrome, 194200<br>Cardiomyopathy, hypertrophic 6, 600858 |
| *PRKCSH* | 99,80% | 95,40% | 100% | 100% | Polycystic liver disease 1, 174050 |
| *PRODH* | 85,00% | 80,60% | 100% | 100% | Hyperprolinemia, type I, 239500 |
| *PRPS1* | 100% | 100% | 100% | 100% | Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070<br>Phosphoribosylpyrophosphate synthetase superactivity, 300661<br>Deafness, X-linked 1, 304500<br>Arts syndrome, 301835<br>Gout, PRPS-related, 300661 |
| *PSAP* | 100% | 100% | 100% | 100% | Gaucher disease, atypical, 610539<br>Krabbe disease, atypical, 611722 |

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|---|---|---|---|---|---|
| | | | | | Combined SAP deficiency, 611721<br>Metachromatic leukodystrophy due to SAP-b deficiency, 249900 |
| *PSAT1* | 95,30% | 81,60% | 100% | 100% | Neu-Laxova syndrome 2, 616038<br>?Phosphoserine aminotransferase deficiency, 610992 |
| *PSPH* | 100% | 100% | 100% | 100% | Phosphoserine phosphatase deficiency, 614023 |
| *PTEN* | 99,50% | 97,00% | 100% | 100% | Prostate cancer, somatic, 176807<br>Cowden syndrome 1, 158350<br>Lhermitte-Duclos syndrome, 158350<br>Macrocephaly/autism syndrome, 605309 |
| *PTGIS* | 98,20% | 95,10% | 100% | 100% | Hypertension, essential, 145500 |
| *PTPN11* | 99,10% | 93,70% | 100% | 100% | LEOPARD syndrome 1, 151100<br>Metachondromatosis, 156250<br>Noonan syndrome 1, 163950<br>Leukemia, juvenile myelomonocytic, somatic, 607785 |
| *PTS* | 99,90% | 99,10% | 100% | 100% | Hyperphenylalaninemia, BH4-deficient, A, 261640 |
| *PUS3* | 100% | 100% | 100% | 100% | Mental retardation, autosomal recessive 55, 617051 |
| *PYCR1* | 99,90% | 97,70% | 100% | 100% | Cutis laxa, autosomal recessive, type IIIB, 614438<br>Cutis laxa, autosomal recessive, type IIB, 612940 |
| *PYCR2* | 100% | 99,10% | 100% | 100% | Leukodystrophy, hypomyelinating, 10, 616420 |
| *PYGL* | 100% | 100% | 100% | 100% | Glycogen storage disease VI, 232700 |
| *PYGM* | 100% | 99,90% | 100% | 100% | McArdle disease, 232600 |
| *QDPR* | 100% | 99,70% | 100% | 100% | Hyperphenylalaninemia, BH4-deficient, C, 261630 |
| *RBCK1* | 99,90% | 98,20% | 100% | 100% | Polyglucosan body myopathy 1 with or without immunodeficiency, 615895 |
| *RDH12* | 100% | 98,60% | 100% | 100% | Leber congenital amaurosis 13, 612712 |
| *RDH5* | 100% | 99,90% | 100% | 100% | Fundus albipunctatus, 136880 |
| *RFT1* | 99,80% | 99,60% | 100% | 100% | Congenital disorder of glycosylation, type In, 612015 |
| *RINT1* | 99,90% | 98,60% | 100% | 100% | Infantile liver failure syndrome 3, 618641 |
| *RPE65* | 99,80% | 97,80% | 100% | 100% | Leber congenital amaurosis 2, 204100<br>Retinitis pigmentosa 87 with choroidal involvement, 618697<br>Retinitis pigmentosa 20, 613794 |
| *RPIA* | 98,60% | 94,90% | 100% | 100% | Ribose 5-phosphate isomerase deficiency, 608611 |
| *SARDH* | 93,70% | 91,70% | 91,40% | 91,40% | No OMIM disease ID |
| *SAT1* | 99,90% | 98,50% | 100% | 99,90% | No OMIM disease ID |
| *SC5D* | 100% | 99,50% | 100% | 100% | Lathosterolosis, 607330 |
| *SCARB2* | 100% | 99,80% | 100% | 100% | Epilepsy, progressive myoclonic 4, with or without renal failure, 254900 |
| *SCP2* | 100% | 99,20% | 100% | 100% | ?Leukoencephalopathy with dystonia and motor neuropathy, 613724 |
| *SCYL1* | 100% | 100% | 100% | 100% | Spinocerebellar ataxia, autosomal recessive 21, 616719 |

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| *SEC23B* | 99,90% | 99,30% | 100% | 100% | ?Cowden syndrome 7, 616858<br>Dyserythropoietic anemia, congenital, type II, 224100 |
| *SELENBP1* | 100% | 100% | 100% | 100% | Extraoral halitosis due to MTO deficiency, 618148 |
| *SEPSECS* | 100% | 100% | 100% | 100% | Pontocerebellar hypoplasia type 2D, 613811 |
| *SERAC1* | 99,90% | 99,50% | 100% | 100% | 3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739 |
| *SGSH* | 94,40% | 94,10% | 100% | 100% | Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900 |
| *SI* | 99,20% | 96,10% | 100% | 100% | Sucrase-isomaltase deficiency, congenital, 222900 |
| *SLC10A7* | 99,70% | 98,00% | 100% | 100% | Short stature, amelogenesis imperfecta, and skeletal dysplasia with scoliosis, 618363 |
| *SLC12A1* | 100% | 99,90% | 100% | 100% | Bartter syndrome, type 1, 601678 |
| *SLC13A3* | 99,40% | 97,50% | 100% | 100% | Leukoencephalopathy, acute reversible, with increased urinary alpha-ketoglutarate, 618384 |
| *SLC16A1* | 100% | 99,30% | 100% | 100% | Monocarboxylate transporter 1 deficiency, 616095<br>Hyperinsulinemic hypoglycemia, familial, 7, 610021<br>Erythrocyte lactate transporter defect, 245340 |
| *SLC17A5* | 99,60% | 97,00% | 100% | 100% | Sialic acid storage disorder, infantile, 269920<br>Salla disease, 604369 |
| *SLC18A2* | 100% | 99,70% | 100% | 100% | ?Parkinsonism-dystonia, infantile, 2, 618049 |
| *SLC22A12* | 100% | 99,80% | 100% | 100% | Hypouricemia, renal, 220150 |
| *SLC22A5* | 100% | 100% | 100% | 100% | Carnitine deficiency, systemic primary, 212140 |
| *SLC25A1* | 95,80% | 88,60% | 99,50% | 97,80% | Myasthenic syndrome, congenital, 23, presynaptic, 618197<br>Combined D-2- and L-2-hydroxyglutaric aciduria, 615182 |
| *SLC25A13* | 100% | 99,70% | 100% | 100% | Citrullinemia, adult-onset type II, 603471<br>Citrullinemia, type II, neonatal-onset, 605814 |
| *SLC25A15* | 99,80% | 98,10% | 100% | 100% | Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970 |
| *SLC25A19* | 100% | 98,50% | 100% | 100% | Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710<br>Microcephaly, Amish type, 607196 |
| *SLC25A20* | 100% | 100% | 100% | 100% | Carnitine-acylcarnitine translocase deficiency, 212138 |
| *SLC25A21* | 100% | 99,70% | 100% | 100% | ?Mitochondrial DNA depletion syndrome 18, 618811 |
| *SLC25A32* | 100% | 100% | 100% | 100% | ?Exercise intolerance, riboflavin-responsive, 616839 |
| *SLC25A38* | 99,70% | 97,10% | 100% | 100% | Anemia, sideroblastic, 2, pyridoxine-refractory, 205950 |
| *SLC25A42* | 96,50% | 93,20% | 100% | 100% | metabolic crises, recurrent, with variable encephalomyopathic features and neurologic regression, 618416 |
| *SLC28A1* | 100% | 98,80% | 100% | 100% | No OMIM disease ID |
| *SLC2A1* | 92,80% | 92,80% | 100% | 100% | Dystonia 9, 601042<br>GLUT1 deficiency syndrome 1, infantile onset, severe, 606777<br>Stomatin-deficient cryohydrocytosis with neurologic defects, 608885<br>GLUT1 deficiency syndrome 2, childhood onset, 612126 |
| *SLC2A2* | 100% | 100% | 100% | 100% | Fanconi-Bickel syndrome, 227810 |

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| *SLC2A9* | 99,80% | 96,10% | 100% | 100% | Hypouricemia, renal, 2, 612076 |
| *SLC30A10* | 100% | 100% | 100% | 100% | Hypermanganesemia with dystonia 1, 613280 |
| *SLC33A1* | 99,90% | 98,90% | 100% | 100% | Spastic paraplegia 42, autosomal dominant, 612539<br>Congenital cataracts, hearing loss, and neurodegeneration, 614482 |
| *SLC35A1* | 100% | 99,70% | 100% | 100% | Congenital disorder of glycosylation, type IIf, 603585 |
| *SLC35A2* | 99,90% | 98,40% | 100% | 100% | Congenital disorder of glycosylation, type IIm, 300896 |
| *SLC35A3* | 80,70% | 78,60% | 81,10% | 81,00% | ?Arthrogryposis, mental retardation, and seizures, 615553 |
| *SLC35C1* | 99,90% | 98,70% | 100% | 100% | Congenital disorder of glycosylation, type IIc, 266265 |
| *SLC35D1* | 100% | 97,70% | 100% | 100% | Schneckenbecken dysplasia, 269250 |
| *SLC37A4* | 100% | 99,20% | 100% | 100% | Glycogen storage disease Ic, 232240<br>Glycogen storage disease Ib, 232220 |
| *SLC39A14* | 100% | 99,40% | 93,50% | 93,50% | ?Hyperostosis cranalis interna, 144755<br>Hypermanganesemia with dystonia 2, 617013 |
| *SLC39A4* | 99,50% | 95,50% | 100% | 100% | Acrodermatitis enteropathica, 201100 |
| *SLC39A8* | 100% | 99,70% | 100% | 100% | Congenital disorder of glycosylation, type IIn, 616721 |
| *SLC3A1* | 100% | 99,80% | 96,60% | 96,60% | Cystinuria, 220100 |
| *SLC44A1* | 98,20% | 98,20% | 100% | 100% | No OMIM disease ID |
| *SLC46A1* | 99,90% | 98,50% | 100% | 100% | Folate malabsorption, hereditary, 229050 |
| *SLC52A1* | 100% | 100% | 100% | 100% | Riboflavin deficiency, 615026 |
| *SLC52A2* | 100% | 100% | 100% | 100% | Brown-Vialetto-Van Laere syndrome 2, 614707 |
| *SLC52A3* | 100% | 100% | 100% | 100% | Brown-Vialetto-Van Laere syndrome 1, 211530<br>?Fazio-Londe disease, 211500 |
| *SLC5A1* | 100% | 100% | 100% | 100% | Glucose/galactose malabsorption, 606824 |
| *SLC5A2* | 100% | 100% | 100% | 100% | Renal glucosuria, 233100 |
| *SLC6A19* | 100% | 100% | 100% | 100% | Iminoglycinuria, digenic, 242600<br>HEARTnup disorder, 234500<br>Hyperglycinuria, 138500 |
| *SLC6A8* | 93,50% | 81,60% | 100% | 99,80% | Cerebral creatine deficiency syndrome 1, 300352 |
| *SLC7A7* | 100% | 99,90% | 100% | 100% | Lysinuric protein intolerance, 222700 |
| *SLC7A9* | 100% | 99,90% | 100% | 100% | Cystinuria, 220100 |
| *SLCO1B1* | 99,20% | 93,70% | 100% | 100% | Hyperbilirubinemia, Rotor type, digenic, 237450 |
| *SLCO1B3* | 98,80% | 90,80% | 100% | 100% | Hyperbilirubinemia, Rotor type, digenic, 237450 |
| *SMPD1* | 100% | 100% | 100% | 100% | Niemann-Pick disease, type A, 257200<br>Niemann-Pick disease, type B, 607616 |
| *SMS* | 91,50% | 78,50% | 100% | 99,90% | Mental retardation, X-linked, Snyder-Robinson type, 309583 |
| *SNX14* | 99,60% | 95,90% | 100% | 100% | Spinocerebellar ataxia, autosomal recessive 20, 616354 |

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| *SOD1* | 100% | 99,90% | 100% | 100% | Amyotrophic lateral sclerosis 1, 105400<br>Spastic tetraplegia and axial hypotonia, progressive, 618598 |
| *SOD2* | 100% | 100% | 100% | 100% | No OMIM disease ID |
| *SPR* | 99,80% | 96,30% | 100% | 100% | Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716 |
| *SPTLC1* | 99,20% | 95,40% | 100% | 100% | Neuropathy, hereditary sensory and autonomic, type IA, 162400 |
| *SPTLC2* | 100% | 100% | 100% | 100% | Neuropathy, hereditary sensory and autonomic, type IC, 613640 |
| *SQOR* | 100% | 97,80% | 100% | 100% | No OMIM disease ID |
| *SRD5A2* | 99,90% | 99,00% | 100% | 100% | Pseudovaginal perineoscrotal hypospadias, 264600 |
| *SRD5A3* | 99,90% | 99,10% | 100% | 100% | Kahrizi syndrome, 612713<br>Congenital disorder of glycosylation, type Iq, 612379 |
| *SSR4* | 100% | 99,70% | 100% | 100% | Congenital disorder of glycosylation, type Iy, 300934 |
| *ST3GAL3* | 100% | 99,80% | 100% | 100% | Mental retardation, autosomal recessive 12, 611090<br>?Epileptic encephalopathy, early infantile, 15, 615006 |
| *ST3GAL5* | 85,00% | 84,20% | 98,70% | 98,40% | Salt and pepper developmental regression syndrome, 609056 |
| *STAR* | 100% | 100% | 100% | 100% | Lipoid adrenal hyperplasia, 201710 |
| *STS* | 99,70% | 98,10% | 100% | 99,90% | Ichthyosis, X-linked, 308100 |
| *STT3A* | 100% | 100% | 100% | 100% | ?Congenital disorder of glycosylation, type Iw, 615596 |
| *STT3B* | 100% | 99,60% | 100% | 100% | ?Congenital disorder of glycosylation, type Ix, 615597 |
| *SUCLA2* | 94,30% | 86,60% | 100% | 100% | Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073 |
| *SUCLG1* | 99,90% | 99,80% | 100% | 100% | Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400 |
| *SUCLG2* | 96,70% | 86,30% | 100% | 100% | No OMIM disease ID |
| *SUGCT* | 99,90% | 98,50% | 100% | 100% | Glutaric aciduria III, 231690 |
| *SUMF1* | 97,50% | 90,80% | 100% | 100% | Multiple sulfatase deficiency, 272200 |
| *SUOX* | 100% | 100% | 100% | 100% | Sulfite oxidase deficiency, 272300 |
| *TBXAS1* | 100% | 100% | 100% | 100% | Ghosal hematodiaphyseal syndrome, 231095 |
| *TCIRG1* | 97,60% | 90,10% | 100% | 100% | Osteopetrosis, autosomal recessive 1, 259700 |
| *TCN2* | 100% | 100% | 100% | 100% | Transcobalamin II deficiency, 275350 |
| *TECR* | 100% | 98,90% | 100% | 100% | Mental retardation, autosomal recessive 14, 614020 |
| *TH* | 99,30% | 96,10% | 100% | 100% | Segawa syndrome, recessive, 605407 |
| *TIMM50* | 98,30% | 94,40% | 100% | 100% | 3-methylglutaconic aciduria, type IX, 617698 |
| *TK2* | 99,20% | 96,30% | 100% | 100% | Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560<br>?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3, 617069 |
| *TKFC* | 100% | 99,50% | 100% | 100% | Triokinase and FMN cyclase deficiency syndrome, 618805 |
| *TKT* | 98,70% | 97,80% | 98,70% | 98,70% | Short stature, developmental delay, and congenital heart defects, 617044 |
| *TMEM165* | 100% | 100% | 100% | 100% | Congenital disorder of glycosylation, type IIk, 614727 |

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| *TMEM199* | 100% | 99,90% | 100% | 100% | Congenital disorder of glycosylation, type IIp, 616829 |
| *TMEM5* | 99,50% | 96,80% | 100% | 99,90% | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041 |
| *TMEM70* | 98,00% | 93,90% | 100% | 100% | Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052 |
| *TMLHE* | 99,50% | 97,10% | 100% | 99,90% | No OMIM disease ID |
| *TPI1* | 99,80% | 97,50% | 100% | 100% | Hemolytic anemia due to triosephosphate isomerase deficiency, 615512 |
| *TPK1* | 99,80% | 99,00% | 100% | 100% | Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458 |
| *TPMT* | 99,10% | 90,10% | 100% | 100% | No OMIM disease ID |
| *TPP1* | 100% | 100% | 100% | 100% | Spinocerebellar ataxia, autosomal recessive 7, 609270<br>Ceroid lipofuscinosis, neuronal, 2, 204500 |
| *TRAK1* | 100% | 99,60% | 100% | 100% | Epileptic encephalopathy, early infantile, 68, 618201 |
| *TRAPPC11* | 100% | 99,20% | 100% | 100% | Muscular dystrophy, limb-girdle, autosomal recessive 18, 615356 |
| *TRAPPC2L* | 100% | 100% | 100% | 100% | Encephalopathy, progressive, early-onset, with episodic rhabdomyolysis, 618331 |
| *TREH* | 96,90% | 92,10% | 100% | 100% | Trehalase deficiency, 612119 |
| *TUSC3* | 100% | 99,50% | 100% | 100% | Mental retardation, autosomal recessive 7, 611093 |
| *TYMP* | 100% | 97,00% | 100% | 100% | Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041 |
| *TYMS* | 99,90% | 99,60% | 100% | 100% | No OMIM disease ID |
| *TYR* | 100% | 100% | 100% | 100% | Waardenburg syndrome/albinism, digenic, 103470<br>Albinism, oculocutaneous, type IB, 606952<br>Albinism, oculocutaneous, type IA, 203100 |
| *TYRP1* | 100% | 99,80% | 100% | 100% | Albinism, oculocutaneous, type III, 203290 |
| *UGT1A1* | 100% | 100% | 100% | 100% | Hyperbilirubinemia, familial transient neonatal, 237900<br>Crigler-Najjar syndrome, type I, 218800<br>Crigler-Najjar syndrome, type II, 606785 |
| *UMPS* | 100% | 99,40% | 97,00% | 97,00% | Orotic aciduria, 258900 |
| *UPB1* | 100% | 100% | 100% | 100% | Beta-ureidopropionase deficiency, 613161 |
| *UROC1* | 100% | 100% | 100% | 100% | ?Urocanase deficiency, 276880 |
| *UROD* | 98,90% | 96,10% | 100% | 100% | Porphyria, hepatoerythropoietic, 176100<br>Porphyria cutanea tarda, 176100 |
| *UROS* | 100% | 99,90% | 100% | 100% | Porphyria, congenital erythropoietic, 263700 |
| *VMA21* | 99,00% | 94,60% | 100% | 98,60% | Myopathy, X-linked, with excessive autophagy, 310440 |
| *VPS13B* | 99,50% | 98,20% | 99,50% | 99,40% | Cohen syndrome, 216550 |
| *VPS33A* | 97,30% | 95,70% | 95,80% | 95,80% | Mucopolysaccharidosis-plus syndrome, 617303 |
| *XDH* | 100% | 99,90% | 100% | 100% | Xanthinuria, type I, 278300 |
| *XYLT1* | 97,40% | 89,60% | 98,10% | 94,80% | Desbuquois dysplasia 2, 615777 |
| *XYLT2* | 100% | 98,30% | 96,70% | 96,70% | Spondyloocular syndrome, 605822 |

*Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan 43(Database issue):D1079-85.*
*Agilent V5 is the default chemistry, and used for all exome analyses apart from the (in-house) TURBO/RAPID WES route.*
*TWIST is the chemistry used for (in-house) TURBO/RAPID WES analysis.*
*Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x.*
*Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x.*
*Genes with coverage denoting NC are non-DNA coding genes.*
*non-DNA coding genes are covered, but as coverage statistics are based on DNA coding regions, statistics could not be generated.*
*OMIM release used for OMIM disease identifiers and descriptions : April 20th , 2020.*
*This list is accurate for panel version DG 2.18*
*Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors*