

Hepatology panel		
versie	v4 (198 genen)	Centrum voor Medische Genetica Gent
Gene	OMIM gene ID	Associated phenotype, OMIM phenotype ID, phenotype mapping key and inheritance pattern
<i>ABCB11</i>	603201	Cholestasis, benign recurrent intrahepatic, 2, 605479 (3), Autosomal recessive; Cholestasis, progressive familial intrahepatic 2, 601847 (3), Autosomal recessive
<i>ABCB4</i>	171060	Gallbladder disease 1, 600803 (3), Autosomal recessive, Autosomal dominant; Cholestasis, intrahepatic, of pregnancy, 3, 614972 (3), Autosomal recessive, Autosomal dominant; Cholestasis, progressive familial intrahepatic 3, 602347 (3), Autosomal recessive
<i>ABCC2</i>	601107	Dubin-Johnson syndrome, 237500 (3), Autosomal recessive
<i>ABCD3</i>	170995	?Bile acid synthesis defect, congenital, 5, 616278 (3), Autosomal recessive
<i>ABCG5</i>	605459	Sitosterolemia 2, 618666 (3), Autosomal recessive
<i>ABCG8</i>	605460	Sitosterolemia 1, 210250 (3), Autosomal recessive; {Gallbladder disease 4}, 611465 (3)
<i>ACADM</i>	607008	Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450 (3), Autosomal recessive
<i>ACADVL</i>	609575	VLCAD deficiency, 201475 (3), Autosomal recessive
<i>ACAT1</i>	607809	Alpha-methylacetoacetic aciduria, 203750 (3), Autosomal recessive
<i>ACOX2</i>	601641	Bile acid synthesis defect, congenital, 6, 617308 (3), Autosomal recessive
<i>ADK</i>	102750	Hypermethioninemia due to adenosine kinase deficiency, 614300 (3), Autosomal recessive
<i>AGL</i>	610860	Glycogen storage disease IIIa, 232400 (3), Autosomal recessive; Glycogen storage disease IIIb, 232400 (3), Autosomal recessive
<i>AGPAT2</i>	603100	Lipodystrophy, congenital generalized, type 1, 608594 (3), Autosomal recessive
<i>AKR1D1</i>	604741	Bile acid synthesis defect, congenital, 2, 235555 (3), Autosomal recessive
<i>ALAD</i>	125270	Porphyria, acute hepatic, 612740 (3), Autosomal recessive; {Lead poisoning, susceptibility to}, 612740 (3), Autosomal recessive
<i>ALDOA</i>	103850	Glycogen storage disease XII, 611881 (3), Autosomal recessive
<i>ALDOB</i>	612724	Fructose intolerance, hereditary, 229600 (3), Autosomal recessive
<i>ALG8</i>	608103	Congenital disorder of glycosylation, type I _h , 608104 (3), Autosomal recessive; Polycystic liver disease 3 with or without kidney cysts, 617874 (3), Autosomal dominant
<i>ALG9</i>	606941	Gillessen-Kaesbach-Nishimura syndrome, 263210 (3), Autosomal recessive; Congenital disorder of glycosylation, type II, 608776 (3), Autosomal recessive

<i>ALMS1</i>	606844	Alstrom syndrome, 203800 (3), Autosomal recessive
<i>AMACR</i>	604489	Alpha-methylacyl-CoA racemase deficiency, 614307 (3), Autosomal recessive; Bile acid synthesis defect, congenital, 4, 214950 (3), Autosomal recessive
<i>ANKS6</i>	615370	Nephronophthisis 16, 615382 (3), Autosomal recessive
<i>AP1S1</i>	603531	MEDNIK syndrome, 609313 (3), Autosomal recessive
<i>ARG1</i>	608313	Argininemia, 207800 (3), Autosomal recessive
<i>ASL</i>	608310	Argininosuccinic aciduria, 207900 (3), Autosomal recessive
<i>ASS1</i>	603470	Citrullinemia, 215700 (3), Autosomal recessive
<i>ATP7B</i>	606882	Wilson disease, 277900 (3), Autosomal recessive
<i>ATP8B1</i>	602397	Cholestasis, progressive familial intrahepatic 1, 211600 (3), Autosomal recessive; Cholestasis, intrahepatic, of pregnancy, 1, 147480 (3), Autosomal dominant; Cholestasis, benign recurrent intrahepatic, 243300 (3), Autosomal recessive
<i>BAAT</i>	602938	Hypercholanemia, familial, 607748 (3), Autosomal recessive; Bile acid conjugation defect 1, 619232 (3), Autosomal recessive
<i>BCS1L</i>	603647	GRACILE syndrome, 603358 (3), Autosomal recessive; Mitochondrial complex III deficiency, nuclear type 1, 124000 (3), Autosomal recessive; Bjornstad syndrome, 262000 (3), Autosomal recessive
<i>BSCL2</i>	606158	Lipodystrophy, congenital generalized, type 2, 269700 (3), Autosomal recessive; Neuropathy, distal hereditary motor, type VC, 619112 (3), Autosomal dominant; Silver spastic paraplegia syndrome, 270685 (3), Autosomal dominant; Encephalopathy, progressive, with or without lipodystrophy, 615924 (3), Autosomal recessive
<i>BTD</i>	609019	Biotinidase deficiency, 253260 (3), Autosomal recessive
<i>CAVIN1</i>	603198	Lipodystrophy, congenital generalized, type 4, 613327 (3), Autosomal recessive
<i>CC2D2A</i>	612013	COACH syndrome 2, 619111 (3); Meckel syndrome 6, 612284 (3), Autosomal recessive; Joubert syndrome 9, 612285 (3), Autosomal recessive
<i>CCDC115</i>	613734	Congenital disorder of glycosylation, type Ilo, 616828 (3), Autosomal recessive
<i>CFTR</i>	602421	Cystic fibrosis, 219700 (3), Autosomal recessive; Sweat chloride elevation without CF (3); Congenital bilateral absence of vas deferens, 277180 (3), Autosomal recessive; {Pancreatitis, hereditary}, 167800 (3), Autosomal dominant; {Bronchiectasis with or without elevated sweat chloride 1, modifier of}, 211400 (3), Autosomal dominant; {Hypertrypsinemia, neonatal} (3)
<i>CLDN1</i>	603718	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis, 607626 (3), Autosomal recessive
<i>COG6</i>	606977	Shaheen syndrome, 615328 (3), Autosomal recessive; Congenital disorder of glycosylation, type III, 614576 (3), Autosomal recessive
<i>COG7</i>	606978	Congenital disorder of glycosylation, type IIe, 608779 (3), Autosomal recessive

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<i>CPS1</i>	608307	Carbamoylphosphate synthetase I deficiency, 237300 (3), Autosomal recessive; {Pulmonary hypertension, neonatal, susceptibility to}, 615371 (3)
<i>CPT1A</i>	600528	CPT deficiency, hepatic, type IA, 255120 (3), Autosomal recessive {Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212 (3), Autosomal recessive, Autosomal dominant; CPT II deficiency, infantile, 600649 (3), Autosomal recessive; CPT II deficiency, lethal neonatal, 608836 (3), Autosomal recessive; CPT II deficiency, myopathic, stress-induced, 255110 (3), Autosomal recessive, Autosomal dominant
<i>CPT2</i>	600650	
<i>CREB3L3</i>	611998	Hypertriglyceridemia 2, 619324 (3), Autosomal dominant
<i>CYP27A1</i>	606530	Cerebrotendinous xanthomatosis, 213700 (3), Autosomal recessive
<i>CYP7A1</i>	118455	No OMIM phenotype
<i>CYP7B1</i>	603711	Spastic paraplegia 5A, autosomal recessive, 270800 (3), Autosomal recessive; Bile acid synthesis defect, congenital, 3, 613812 (3), Autosomal recessive
<i>DCDC2</i>	605755	Nephronophthisis 19, 616217 (3), Autosomal recessive; ?Deafness, autosomal recessive 66, 610212 (3), Autosomal recessive; Sclerosing cholangitis, neonatal, 617394 (3), Autosomal recessive
<i>DGUOK</i>	601465	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, 617070 (3), Autosomal recessive; Portal hypertension, noncirrhotic, 617068 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880 (3), Autosomal recessive
<i>DHCR7</i>	602858	Smith-Lemli-Opitz syndrome, 270400 (3), Autosomal recessive
<i>DKC1</i>	300126	Dyskeratosis congenita, X-linked, 305000 (3), X-linked recessive
<i>DLD</i>	238331	Dihydrolipoamide dehydrogenase deficiency, 246900 (3), Autosomal recessive
<i>DNAJB11</i>	611341	Polycystic kidney disease 6 with or without polycystic liver disease, 618061 (3), Autosomal dominant
<i>EHHADH</i>	607037	?Fanconi renal tubular syndrome 3, 615605 (3), Autosomal dominant
<i>EIF2AK3</i>	604032	Wolcott-Rallison syndrome, 226980 (3), Autosomal recessive
<i>ENO3</i>	131370	?Glycogen storage disease XIII, 612932 (3), Autosomal recessive
<i>EPHX1</i>	132810	No OMIM phenotype
<i>EPM2A</i>	607566	Epilepsy, progressive myoclonic 2A (Lafora), 254780 (3), Autosomal recessive
<i>ETFA</i>	608053	Glutaric acidemia IIA, 231680 (3), Autosomal recessive
<i>ETFB</i>	130410	Glutaric acidemia IIB, 231680 (3), Autosomal recessive
<i>FAH</i>	613871	Tyrosinemia, type I, 276700 (3), Autosomal recessive
<i>FARSA</i>	602918	?Rajab interstitial lung disease with brain calcifications 2, 619013 (3), Autosomal recessive
<i>FARSB</i>	609690	Rajab interstitial lung disease with brain calcifications 1, 613658 (3), Autosomal recessive
<i>FBP1</i>	611570	Fructose-1,6-bisphosphatase deficiency, 229700 (3), Autosomal recessive
<i>FTH1</i>	134770	?Hemochromatosis, type 5, 615517 (3), Autosomal dominant

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<i>G6PC1</i>	613742	Glycogen storage disease Ia, 232200 (3), Autosomal recessive
<i>GAA</i>	606800	Glycogen storage disease II, 232300 (3), Autosomal recessive
<i>GALE</i>	606953	Galactose epimerase deficiency, 230350 (3), Autosomal recessive
<i>GALK1</i>	604313	Galactokinase deficiency with cataracts, 230200 (3), Autosomal recessive
<i>GALM</i>	137030	Galactosemia IV, 618881 (3), Autosomal recessive
<i>GALT</i>	606999	Galactosemia, 230400 (3), Autosomal recessive
<i>GANAB</i>	104160	Polycystic kidney disease 3, 600666 (3), Autosomal dominant
<i>GBA</i>	606463	{Lewy body dementia, susceptibility to}, 127750 (3), Autosomal dominant; Gaucher disease, type II, 230900 (3), Autosomal recessive; Gaucher disease, type IIIC, 231005 (3), Autosomal recessive; Gaucher disease, type III, 231000 (3), Autosomal recessive; Gaucher disease, type I, 230800 (3), Autosomal recessive; Gaucher disease, perinatal lethal, 608013 (3), Autosomal recessive; {Parkinson disease, late-onset, susceptibility to}, 168600 (3), Multifactorial, Autosomal dominant
<i>GBE1</i>	607839	Glycogen storage disease IV, 232500 (3), Autosomal recessive; Polyglucosan body disease, adult form, 263570 (3), Autosomal recessive
<i>GFM1</i>	606639	Combined oxidative phosphorylation deficiency 1, 609060 (3), Autosomal recessive
<i>GNAS</i>	139320	ACTH-independent macronodular adrenal hyperplasia, 219080 (3), Somatic mutation; Pituitary adenoma 3, multiple types, somatic, 617686 (3); Pseudohypoparathyroidism Ic, 612462 (3), Autosomal dominant; Pseudohypoparathyroidism Ia, 103580 (3), Autosomal dominant; Osseous heteroplasia, progressive, 166350 (3), Autosomal dominant; Pseudohypoparathyroidism Ib, 603233 (3), Autosomal dominant; McCune-Albright syndrome, somatic, mosaic, 174800 (3); Pseudopseudohypoparathyroidism, 612463 (3), Autosomal dominant
<i>GUSB</i>	611499	Mucopolysaccharidosis VII, 253220 (3), Autosomal recessive
<i>GYS1</i>	138570	Glycogen storage disease 0, muscle, 611556 (3), Autosomal recessive
<i>GYS2</i>	138571	Glycogen storage disease 0, liver, 240600 (3), Autosomal recessive
<i>HADHA</i>	600890	HELLP syndrome, maternal, of pregnancy, 609016 (3), Autosomal recessive; Mitochondrial trifunctional protein deficiency, 609015 (3), Autosomal recessive; LCHAD deficiency, 609016 (3), Autosomal recessive; Fatty liver, acute, of pregnancy, 609016 (3), Autosomal recessive
<i>HAMP</i>	606464	Hemochromatosis, type 2B, 613313 (3), Autosomal recessive
<i>HFE</i>	613609	{Porphyria variegata, susceptibility to}, 176200 (3), Autosomal dominant; {Microvascular complications of diabetes 7}, 612635 (3); Hemochromatosis, 235200 (3), Autosomal recessive; {Alzheimer disease, susceptibility to}, 104300 (3), Autosomal dominant; [Transferrin serum level QTL2], 614193 (3); {Porphyria cutanea tarda, susceptibility to}, 176100 (3), Autosomal recessive, Autosomal dominant
<i>HJV</i>	608374	Hemochromatosis, type 2A, 602390 (3), Autosomal recessive

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<i>HLCS</i>	609018	Holocarboxylase synthetase deficiency, 253270 (3), Autosomal recessive
<i>HNF1B</i>	189907	Type 2 diabetes mellitus, 125853 (3), Autosomal dominant; Renal cysts and diabetes syndrome, 137920 (3), Autosomal dominant; {Renal cell carcinoma}, 144700 (3)
<i>HSD17B4</i>	601860	D-bifunctional protein deficiency, 261515 (3), Autosomal recessive; Perrault syndrome 1, 233400 (3), Autosomal recessive
<i>HSD3B7</i>	607764	Bile acid synthesis defect, congenital, 1, 607765 (3), Autosomal recessive
<i>IARS1</i>	600709	Growth retardation, impaired intellectual development, hypotonia, and hepatopathy, 617093 (3), Autosomal recessive
<i>IFT140</i>	614620	Short-rib thoracic dysplasia 9 with or without polydactyly, 266920 (3), Autosomal recessive; Retinitis pigmentosa 80, 617781 (3), Autosomal recessive
<i>INVS</i>	243305	Nephronophthisis 2, infantile, 602088 (3), Autosomal recessive
<i>IVD</i>	607036	Isovaleric acidemia, 243500 (3), Autosomal recessive
<i>JAG1</i>	601920	?Deafness, congenital heart defects, and posterior embryotoxon, 617992 (3), Autosomal dominant; Alagille syndrome 1, 118450 (3), Autosomal dominant; Tetralogy of Fallot, 187500 (3), Autosomal dominant
<i>KIF12</i>	611278	Cholestasis, progressive familial intrahepatic, 8, 619662 (3), Autosomal recessive
<i>KRT18</i>	148070	Cirrhosis, cryptogenic, 215600 (3), Autosomal recessive; {Cirrhosis, noncryptogenic, susceptibility to}, 215600 (3), Autosomal recessive
<i>KRT8</i>	148060	{Cirrhosis, noncryptogenic, susceptibility to}, 215600 (3), Autosomal recessive; Cirrhosis, cryptogenic, 215600 (3), Autosomal recessive
<i>LAMP2</i>	309060	Danon disease, 300257 (3), X-linked dominant
<i>LARS1</i>	151350	?Infantile liver failure syndrome 1, 615438 (3), Autosomal recessive
<i>LDHA</i>	150000	Glycogen storage disease XI, 612933 (3), Autosomal recessive
<i>LIPA</i>	613497	Wolman disease, 278000 (3), Autosomal recessive; Cholesteryl ester storage disease, 278000 (3), Autosomal recessive
<i>LRP5</i>	603506	Osteopetrosis, autosomal dominant 1, 607634 (3), Autosomal dominant; [Bone mineral density variability 1], 601884 (3), Autosomal dominant; {Osteoporosis}, 166710 (3), Autosomal dominant; Hyperostosis, endosteal, 144750 (3), Autosomal dominant; Osteosclerosis, 144750 (3), Autosomal dominant; Polycystic liver disease 4 with or without kidney cysts, 617875 (3), Autosomal dominant; Osteoporosis-pseudoglioma syndrome, 259770 (3), Autosomal recessive; Exudative vitreoretinopathy 4, 601813 (3), Autosomal recessive, Autosomal dominant; van Buchem disease, type 2, 607636 (3)
<i>LSR</i>	616582	No OMIM phenotype
<i>MARS1</i>	156560	Interstitial lung and liver disease, 615486 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal, type 2U, 616280 (3), Autosomal dominant

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<i>MCEE</i>	608419	Methylmalonyl-CoA epimerase deficiency, 251120 (3), Autosomal recessive
<i>MKS1</i>	609883	Bardet-Biedl syndrome 13, 615990 (3), Autosomal recessive; Meckel syndrome 1, 249000 (3), Autosomal recessive; Joubert syndrome 28, 617121 (3), Autosomal recessive
<i>MMUT</i>	609058	Methylmalonic aciduria, mut(0) type, 251000 (3), Autosomal recessive
<i>MPI</i>	154550	Congenital disorder of glycosylation, type Ib, 602579 (3), Autosomal recessive
<i>MPV17</i>	137960	Charcot-Marie-Tooth disease, axonal, type 2EE, 618400 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810 (3), Autosomal recessive
<i>MVK</i>	251170	Hyper-IgD syndrome, 260920 (3), Autosomal recessive; Porokeratosis 3, multiple types, 175900 (3), Autosomal dominant; Mevalonic aciduria, 610377 (3), Autosomal recessive
<i>MYO5B</i>	606540	Diarrhea 2, with microvillus atrophy, 251850 (3), Autosomal recessive
<i>NBAS</i>	608025	Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800 (3), Autosomal recessive; Infantile liver failure syndrome 2, 616483 (3), Autosomal recessive
<i>NEK8</i>	609799	Renal-hepatic-pancreatic dysplasia 2, 615415 (3), Autosomal recessive; ?Nephronophthisis 9, 613824 (3)
<i>NEU1</i>	608272	Sialidosis, type II, 256550 (3), Autosomal recessive; Sialidosis, type I, 256550 (3), Autosomal recessive
<i>NGLY1</i>	610661	Congenital disorder of deglycosylation, 615273 (3), Autosomal recessive
<i>NHLRC1</i>	608072	Epilepsy, progressive myoclonic 2B (Lafora), 254780 (3), Autosomal recessive
<i>NOTCH2</i>	600275	Alagille syndrome 2, 610205 (3), Autosomal dominant; Hajdu-Cheney syndrome, 102500 (3), Autosomal dominant
<i>NPC1</i>	607623	Niemann-Pick disease, type C1, 257220 (3), Autosomal recessive; Niemann-Pick disease, type D, 257220 (3), Autosomal recessive
<i>NPC2</i>	601015	Niemann-pick disease, type C2, 607625 (3), Autosomal recessive
<i>NPHP1</i>	607100	Joubert syndrome 4, 609583 (3), Autosomal recessive; Nephronophthisis 1, juvenile, 256100 (3), Autosomal recessive; Senior-Loken syndrome-1, 266900 (3), Autosomal recessive
<i>NPHP3</i>	608002	Nephronophthisis 3, 604387 (3), Autosomal recessive; Renal-hepatic-pancreatic dysplasia 1, 208540 (3), Autosomal recessive; Meckel syndrome 7, 267010 (3), Autosomal recessive
<i>NPHP4</i>	607215	Senior-Loken syndrome 4, 606996 (3), Autosomal recessive; Nephronophthisis 4, 606966 (3), Autosomal recessive
<i>NR1H4</i>	603826	Cholestasis, progressive familial intrahepatic, 5, 617049 (3), Autosomal recessive
<i>OXCT1</i>	601424	Succinyl CoA:3-oxoacid CoA transferase deficiency, 245050 (3), Autosomal recessive
<i>PC</i>	608786	Pyruvate carboxylase deficiency, 266150 (3), Autosomal recessive
<i>PCCA</i>	232000	Propionicacidemia, 606054 (3), Autosomal recessive

<i>PCCB</i>	232050	Propionicacidemia, 606054 (3), Autosomal recessive
<i>PEX1</i>	602136	Heimler syndrome 1, 234580 (3), Autosomal recessive; Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 (3), Autosomal recessive; Peroxisome biogenesis disorder 1A (Zellweger), 214100 (3), Autosomal recessive
<i>PEX10</i>	602859	Peroxisome biogenesis disorder 6A (Zellweger), 614870 (3), Autosomal recessive; Peroxisome biogenesis disorder 6B, 614871 (3), Autosomal recessive
<i>PEX11B</i>	603867	Peroxisome biogenesis disorder 14B, 614920 (3), Autosomal recessive
<i>PEX12</i>	601758	Peroxisome biogenesis disorder 3B, 266510 (3), Autosomal recessive; Peroxisome biogenesis disorder 3A (Zellweger), 614859 (3), Autosomal recessive
<i>PEX13</i>	601789	Peroxisome biogenesis disorder 11A (Zellweger), 614883 (3), Autosomal recessive; Peroxisome biogenesis disorder 11B, 614885 (3), Autosomal recessive
<i>PEX14</i>	601791	Peroxisome biogenesis disorder 13A (Zellweger), 614887 (3), Autosomal recessive
<i>PEX16</i>	603360	Peroxisome biogenesis disorder 8B, 614877 (3), Autosomal recessive; Peroxisome biogenesis disorder 8A (Zellweger), 614876 (3), Autosomal recessive
<i>PEX19</i>	600279	Peroxisome biogenesis disorder 12A (Zellweger), 614886 (3), Autosomal recessive
<i>PEX2</i>	170993	Peroxisome biogenesis disorder 5A (Zellweger), 614866 (3), Autosomal recessive; Peroxisome biogenesis disorder 5B, 614867 (3), Autosomal recessive
<i>PEX26</i>	608666	Peroxisome biogenesis disorder 7B, 614873 (3), Autosomal recessive; Peroxisome biogenesis disorder 7A (Zellweger), 614872 (3), Autosomal recessive
<i>PEX3</i>	603164	Peroxisome biogenesis disorder 10A (Zellweger), 614882 (3), Autosomal recessive; ?Peroxisome biogenesis disorder 10B, 617370 (3), Autosomal recessive
<i>PEX5</i>	600414	Peroxisome biogenesis disorder 2B, 202370 (3), Autosomal recessive; Peroxisome biogenesis disorder 2A (Zellweger), 214110 (3), Autosomal recessive; Rhizomelic chondrodysplasia punctata, type 5, 616716 (3), Autosomal recessive
<i>PEX6</i>	601498	Peroxisome biogenesis disorder 4B, 614863 (3), Autosomal recessive, Autosomal dominant; Peroxisome biogenesis disorder 4A (Zellweger), 614862 (3), Autosomal recessive; Heimler syndrome 2, 616617 (3), Autosomal recessive
<i>PEX7</i>	601757	Rhizomelic chondrodysplasia punctata, type 1, 215100 (3), Autosomal recessive; Peroxisome biogenesis disorder 9B, 614879 (3), Autosomal recessive
<i>PFKM</i>	610681	Glycogen storage disease VII, 232800 (3), Autosomal recessive
<i>PGAM2</i>	612931	Glycogen storage disease X, 261670 (3), Autosomal recessive

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<i>PGK1</i>	311800	Phosphoglycerate kinase 1 deficiency, 300653 (3), X-linked recessive
<i>PGM1</i>	171900	Congenital disorder of glycosylation, type It, 614921 (3), Autosomal recessive
<i>PHKA1</i>	311870	Muscle glycogenosis, 300559 (3), X-linked recessive
<i>PHKA2</i>	300798	Glycogen storage disease, type IXa2, 306000 (3), X-linked recessive; Glycogen storage disease, type IXa1, 306000 (3), X-linked recessive
<i>PHKB</i>	172490	Phosphorylase kinase deficiency of liver and muscle, autosomal recessive, 261750 (3), Autosomal recessive
<i>PHKG2</i>	172471	Glycogen storage disease IXc, 613027 (3), Autosomal recessive
<i>PKD1</i>	601313	Polycystic kidney disease 1, 173900 (3), Autosomal dominant
<i>PKD2</i>	173910	Polycystic kidney disease 2, 613095 (3), Autosomal dominant
<i>PKHD1</i>	606702	Polycystic kidney disease 4, with or without hepatic disease, 263200 (3), Autosomal recessive
<i>PMM2</i>	601785	Congenital disorder of glycosylation, type Ia, 212065 (3), Autosomal recessive
<i>PNPLA3</i>	609567	No OMIM phenotype
<i>POLG</i>	174763	Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 (3), Autosomal recessive; Progressive external ophthalmoplegia, autosomal dominant 1, 157640 (3), Autosomal dominant; Progressive external ophthalmoplegia, autosomal recessive 1, 258450 (3), Autosomal recessive
<i>PPM1F</i>	619309	No OMIM phenotype
<i>PRKAG2</i>	602743	Glycogen storage disease of heart, lethal congenital, 261740 (3), Autosomal dominant; Wolff-Parkinson-White syndrome, 194200 (3), Autosomal dominant; Cardiomyopathy, hypertrophic 6, 600858 (3), Autosomal dominant
<i>PRKCSH</i>	177060	Polycystic liver disease 1, 174050 (3), Autosomal dominant
<i>PYGL</i>	613741	Glycogen storage disease VI, 232700 (3), Autosomal recessive
<i>PYGM</i>	608455	McArdle disease, 232600 (3), Autosomal recessive
<i>RBCK1</i>	610924	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895 (3), Autosomal recessive
<i>RINT1</i>	610089	Infantile liver failure syndrome 3, 618641 (3), Autosomal recessive
<i>RPGRIP1L</i>	610937	Joubert syndrome 7, 611560 (3), Autosomal recessive; Meckel syndrome 5, 611561 (3), Autosomal recessive; ?COACH syndrome 3, 619113 (3)
<i>SBDS</i>	607444	{Aplastic anemia, susceptibility to}, 609135 (3); Shwachman-Diamond syndrome, 260400 (3), Autosomal recessive
<i>SCO1</i>	603644	Mitochondrial complex IV deficiency, nuclear type 4, 619048 (3), Autosomal recessive
<i>SEC63</i>	608648	Polycystic liver disease 2, 617004 (3), Autosomal dominant

<i>SERPINA1</i>	107400	Hemorrhagic diathesis due to antithrombin Pittsburgh, 613490 (3), Autosomal recessive; Emphysema due to AAT deficiency, 613490 (3), Autosomal recessive; Emphysema-cirrhosis, due to AAT deficiency, 613490 (3), Autosomal recessive
<i>SLC10A1</i>	182396	Hypercholanemia, familial 2, 619256 (3), Autosomal recessive
<i>SLC10A2</i>	601295	Bile acid malabsorption, primary, 613291 (3), Autosomal recessive
<i>SLC16A1</i>	600682	Hyperinsulinemic hypoglycemia, familial, 7, 610021 (3), Autosomal dominant; Erythrocyte lactate transporter defect, 245340 (3), Autosomal dominant; Monocarboxylate transporter 1 deficiency, 616095 (3), Autosomal recessive, Autosomal dominant
<i>SLC25A13</i>	603859	Citrullinemia, type II, neonatal-onset, 605814 (3), Autosomal recessive; Citrullinemia, adult-onset type II, 603471 (3), Autosomal recessive
<i>SLC25A20</i>	613698	Carnitine-acylcarnitine translocase deficiency, 212138 (3), Autosomal recessive
<i>SLC2A2</i>	138160	Fanconi-Bickel syndrome, 227810 (3), Autosomal recessive; {Diabetes mellitus, noninsulin-dependent}, 125853 (3), Autosomal dominant
<i>SLC30A10</i>	611146	Hypermanganesemia with dystonia 1, 613280 (3), Autosomal recessive
<i>SLC51A</i>	612084	?Cholestasis, progressive familial intrahepatic, 6, 619484 (3), Autosomal recessive
<i>SLCO1B1</i>	604843	Hyperbilirubinemia, Rotor type, digenic, 237450 (3), Digenic recessive
<i>SLCO1B3</i>	605495	Hyperbilirubinemia, Rotor type, digenic, 237450 (3), Digenic recessive
<i>SMPD1</i>	607608	Niemann-Pick disease, type B, 607616 (3), Autosomal recessive; Niemann-Pick disease, type A, 257200 (3), Autosomal recessive
<i>SP110</i>	604457	{Mycobacterium tuberculosis, susceptibility to}, 607948 (3); Hepatic venoocclusive disease with immunodeficiency, 235550 (3), Autosomal recessive
<i>STN1</i>	613128	Cerebroretinal microangiopathy with calcifications and cysts 2, 617341 (3), Autosomal recessive
<i>STT3B</i>	608605	?Congenital disorder of glycosylation, type Ix, 615597 (3), Autosomal recessive
<i>TALDO1</i>	602063	Transaldolase deficiency, 606003 (3), Autosomal recessive
<i>TANGO2</i>	616830	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878 (3), Autosomal recessive
<i>TFR2</i>	604720	Hemochromatosis, type 3, 604250 (3), Autosomal recessive
<i>TJP2</i>	607709	Hypercholanemia, familial 1, 607748 (3), Autosomal recessive; Cholestasis, progressive familial intrahepatic 4, 615878 (3), Autosomal recessive
<i>TMEM216</i>	613277	Joubert syndrome 2, 608091 (3), Autosomal recessive; Meckel syndrome 2, 603194 (3), Autosomal recessive

<i>TMEM67</i>	609884	Nephronophthisis 11, 613550 (3), Autosomal recessive; {Bardet-Biedl syndrome 14, modifier of}, 615991 (3), Autosomal recessive; Joubert syndrome 6, 610688 (3), Autosomal recessive; Meckel syndrome 3, 607361 (3), Autosomal recessive; ?RHYNS syndrome, 602152 (3), Autosomal recessive; COACH syndrome 1, 216360 (3), Autosomal recessive
<i>TRMU</i>	610230	{Deafness, mitochondrial, modifier of}, 580000 (3), Mitochondrial; Liver failure, transient infantile, 613070 (3), Autosomal recessive
<i>TTC37</i>	614589	Trichohepatoenteric syndrome 1, 222470 (3), Autosomal recessive
<i>TWNK</i>	606075	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 (3), Autosomal recessive; Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286 (3), Autosomal dominant; Perrault syndrome 5, 616138 (3), Autosomal recessive
<i>UGT1A1</i>	191740	Crigler-Najjar syndrome, type I, 218800 (3), Autosomal recessive; [Bilirubin, serum level of, QTL1], 601816 (3); Hyperbilirubinemia, familial transient neonatal, 237900 (3), Autosomal recessive; Crigler-Najjar syndrome, type II, 606785 (3), Autosomal recessive; [Gilbert syndrome], 143500 (3), Autosomal recessive
<i>UNC45A</i>	611219	Osteotohepatoenteric syndrome, 619377 (3), Autosomal recessive
<i>UROS</i>	606938	Porphyria, congenital erythropoietic, 263700 (3), Autosomal recessive
<i>USP53</i>	617431	Cholestasis, progressive familial intrahepatic, 7, with or without hearing loss, 619658 (3), Autosomal recessive
<i>UTP4</i>	607456/604901	-/North American Indian childhood cirrhosis, 604901 (2)
<i>VIPAS39</i>	613401	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404 (3), Autosomal recessive
<i>VPS33B</i>	608552	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085 (3), Autosomal recessive
<i>VPS50</i>	616465	Neurodevelopmental disorder with microcephaly, seizures, and neonatal cholestasis, 619685 (3), Autosomal recessive
<i>WDR83OS</i>	618474	No OMIM phenotype
<i>YARS1</i>	603623	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease 2, 619418 (3), Autosomal recessive; Charcot-Marie-Tooth disease, dominant intermediate C, 608323 (3), Autosomal dominant
<i>ZFYVE19</i>	619635	Cholestasis, progressive familial intrahepatic, 9, 619849 (3), Autosomal recessive

Gene symbols used are according to the HGNC guidelines. For some genes a previously HGNC-approved symbol is in brackets.

Each Phenotype is followed by its MIM number, phenotype mapping key and inheritance pattern.

OMIM release used for OMIM disease identifiers and descriptions: July 26, 2021

Possible phenotype mapping keys

- (1) the disorder is placed on the map based on its association with a gene, but the underlying defect is not known
- (2) the disorder has been placed on the map by linkage; no mutation has been found
- (3) the molecular basis for the disorder is known; a mutation has been found in the gene
- (4) a contiguous gene deletion or duplication syndrome, multiple genes are deleted or duplicated causing the phenotype

Brackets, "[]", indicate "nondiseases," mainly genetic variations that lead to apparently abnormal laboratory test values (e.g., dysalbuminemic euthyroidal hyperthyroxinemia).

Braces, "{ }", indicate mutations that contribute to susceptibility to multifactorial disorders (e.g., diabetes, asthma) or to susceptibility to infection (e.g., malaria).

A question mark, "?", before the phenotype name indicates that the relationship between the phenotype and gene is provisional. More details about this relationship are provided in the comment field of the map and in the gene and phenotype OMIM entries.