Hepatology

Gene panel

Gene panel information

Gene panel	**Hepatology**
Version	6
Total genes	254
Activation date	Friday 21 june 2024
Publisher	Center for Medical Genetics, Ghent

Genes

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
ABCB11	99.86 %	603201	Cholestasis, benign recurrent intrahepatic, 2, 605479 (3), Autosomal recessive; Cholestasis, progressive familial intrahepatic 2, 601847 (3), Autosomal recessive
ABCB4	99.71 %	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
ABCC2	99.94 %	601107	Dubin-Johnson syndrome, 237500 (3), Autosomal recessive
ABCD3	92.7 %	170995	?Bile acid synthesis defect, congenital, 5, 616278 (3), Autosomal recessive
ABCG5	99.96 %	605459	Sitosterolemia 2, 618666 (3), Autosomal recessive
ABCG8	99.95 %	605460	Sitosterolemia 1, 210250 (3), Autosomal recessive; {Gallbladder disease 4}, 611465 (3)
ACADM	96.14 %	607008	Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450 (3), Autosomal recessive
ACADVL	100 %	609575	VLCAD deficiency, 201475 (3), Autosomal recessive
ACAT1	99.81 %	607809	Alpha-methylacetoacetic aciduria, 203750 (3), Autosomal recessive
ACOX2	99.81 %	601641	Bile acid synthesis defect, congenital, 6, 617308 (3), Autosomal recessive
ACVRL1	99.88 %	601284	Telangiectasia, hereditary hemorrhagic, type 2, 600376 (3), Autosomal dominant
ADK	99.78 %	102750	Hypermethioninemia due to adenosine kinase deficiency, 614300 (3), Autosomal recessive
AGL	97.67 %	610860	Glycogen storage disease IIIa, 232400 (3), Autosomal recessive; Glycogen storage disease IIIb, 232400 (3), Autosomal recessive
AGPAT2	100 %	603100	Lipodystrophy, congenital generalized, type 1, 608594 (3), Autosomal recessive
AGXT	100 %	604285	Hyperoxaluria, primary, type 1, 259900 (3), Autosomal recessive
AKR1D1	99.91 %	604741	Bile acid synthesis defect, congenital, 2, 235555 (3), Autosomal recessive
ALAD	99.99 %	125270	Porphyria, acute hepatic, 612740 (3), Autosomal recessive; {Lead poisoning, susceptibility to}, 612740 (3), Autosomal recessive
ALAS2	99.98 %	301300	Anemia, sideroblastic, 1, 300751 (3), X-linked recessive; Protoporphyria, erythropoietic, X-linked, 300752 (3), X-linked
ALDOA	100 %	103850	Glycogen storage disease XII, 611881 (3), Autosomal recessive
ALDOB	100 %	612724	Fructose intolerance, hereditary, 229600 (3), Autosomal recessive
ALG8	95.49 %	608103	Congenital disorder of glycosylation, type Ih, 608104 (3), Autosomal recessive; Polycystic liver disease 3 with or without kidney cysts, 617874 (3), Autosomal dominant
ALG9	99.73 %	606941	Gillessen-Kaesbach-Nishimura syndrome, 263210 (3), Autosomal recessive; Congenital disorder of glycosylation, type Il, 608776 (3), Autosomal recessive
ALMS1	99.9 %	606844	Alstrom syndrome, 203800 (3), Autosomal recessive

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AMACR	100 %	604489	Alpha-methylacyl-CoA racemase deficiency, 614307 (3), Autosomal recessive; Bile acid synthesis defect, congenital, 4, 214950 (3), Autosomal recessive
ANKS6	100 %	615370	Nephronophthisis 16, 615382 (3), Autosomal recessive
AP1S1	99.49 %	603531	MEDNIK syndrome, 609313 (3), Autosomal recessive
ARG1	99.95 %	608313	Argininemia, 207800 (3), Autosomal recessive
ASL	99.98 %	608310	Argininosuccinic aciduria, 207900 (3), Autosomal recessive
ASS1	77.52 %	603470	Citrullinemia, 215700 (3), Autosomal recessive
ATP11C	99.66 %	300516	?Hemolytic anemia, congenital, X-linked, 301015 (3), X-linked recessive
ATP6AP1	100 %	300197	Immunodeficiency 47, 300972 (3), X-linked recessive
ATP7B	100 %	606882	Wilson disease, 277900 (3), Autosomal recessive
ATP8B1	99.94 %	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
BAAT	99.98 %	602938	Bile acid conjugation defect 1, 619232 (3), Autosomal recessive
BCS1L	99.99 %	603647	GRACILE syndrome, 603358 (3), Autosomal recessive; Mitochondrial complex III deficiency, nuclear type 1, 124000 (3), Autosomal recessive; Bjornstad syndrome, 262000 (3), Autosomal recessive
BMP6	100 %	112266	{Iron overload, susceptibility to}, 620121 (3), Autosomal dominant
BOLA3	99.22 %	613183	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299 (3), Autosomal recessive
BSCL2	99.99 %	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
BTD	100 %	609019	Biotinidase deficiency, 253260 (3), Autosomal recessive
CACNA1E	99.82 %	601013	Developmental and epileptic encephalopathy 69, 618285 (3), Autosomal dominant
CAVIN1	100 %	603198	Lipodystrophy, congenital generalized, type 4, 613327 (3), Autosomal recessive
CC2D2A	99.95 %	612013	COACH syndrome 2, 619111 (3), Autosomal recessive; Retinitis pigmentosa 93, 619845 (3), Autosomal recessive; Meckel syndrome 6, 612284 (3), Autosomal recessive; Joubert syndrome 9, 612285 (3), Autosomal recessive
CCDC115	99.9 %	613734	Congenital disorder of glycosylation, type IIo, 616828 (3), Autosomal recessive
CCDC88B	99.99 %	611205	*No OMIM phenotypes*
CDAN1	100 %	607465	Dyserythropoietic anemia, congenital, type Ia, 224120 (3), Autosomal recessive
CFTR	99.45 %	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)
CLDN1	99.99 %	603718	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis, 607626 (3), Autosomal recessive
COG6	99.86 %	606977	Shaheen syndrome, 615328 (3), Autosomal recessive; Congenital disorder of glycosylation, type III, 614576 (3), Autosomal recessive
COG7	99.74 %	606978	Congenital disorder of glycosylation, type IIe, 608779 (3), Autosomal recessive
CP	99.95 %	117700	Cerebellar ataxia, 604290 (3), Autosomal recessive; [Hypoceruloplasminemia, hereditary], 604290 (3), Autosomal recessive; Hemosiderosis, systemic, due to aceruloplasminemia, 604290 (3), Autosomal recessive
CPS1	99.91 %	608307	Carbamoylphosphate synthetase I deficiency, 237300 (3), Autosomal recessive; {Pulmonary hypertension, neonatal, susceptibility to}, 615371 (3)

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CPT1A	99.98 %	600528	CPT deficiency, hepatic, type IA, 255120 (3), Autosomal recessive
CPT2	99.65 %	600650	{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
CREB3L3	99.98 %	611998	Hypertriglyceridemia 2, 619324 (3), Autosomal dominant
CYP27A1	100 %	606530	Cerebrotendinous xanthomatosis, 213700 (3), Autosomal recessive
CYP7A1	99.99 %	118455	*No OMIM phenotypes*
CYP7B1	99.82 %	603711	Spastic paraplegia 5A, autosomal recessive, 270800 (3), Autosomal recessive; Bile acid synthesis defect, congenital, 3, 613812 (3), Autosomal recessive
DCDC2	99.96 %	605755	Nephronophthisis 19, 616217 (3), Autosomal recessive; ?Deafness, autosomal recessive 66, 610212 (3), Autosomal recessive; Sclerosing cholangitis, neonatal, 617394 (3), Autosomal recessive
DGUOK	99.93 %	601465	Portal hypertension, noncirrhotic, 1, 617068 (3), Autosomal recessive; Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, 617070 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880 (3), Autosomal recessive
DHCR7	99.97 %	602858	Smith-Lemli-Opitz syndrome, 270400 (3), Autosomal recessive
DKC1	99.59 %	300126	?Cataracts, hearing impairment, nephrotic syndrome, and enterocolitis 1, 301108 (3), X-linked dominant; Dyskeratosis congenita, X-linked, 305000 (3), X-linked recessive
DLD	99.89 %	238331	Dihydrolipoamide dehydrogenase deficiency, 246900 (3), Autosomal recessive
DNAJB11	99.97 %	611341	Polycystic kidney disease 6 with or without polycystic liver disease, 618061 (3), Autosomal dominant
DNAJC21	99.67 %	617048	Bone marrow failure syndrome 3, 617052 (3), Autosomal recessive
EFL1	99.83 %	617538	Shwachman-Diamond syndrome 2, 617941 (3), Autosomal recessive
EHHADH	99.99 %	607037	?Fanconi renotubular syndrome 3, 615605 (3), Autosomal dominant
EIF2AK3	97.43 %	604032	Wolcott-Rallison syndrome, 226980 (3), Autosomal recessive
ENO3	100 %	131370	Glycogen storage disease XIII, 612932 (3), Autosomal recessive
EPB41	97.07 %	130500	Elliptocytosis-1, 611804 (3), Autosomal recessive, Autosomal dominant
EPHX1	99.97 %	132810	*No OMIM phenotypes*
EPM2A	99.99 %	607566	Epilepsy, progressive myoclonic 2A (Lafora), 254780 (3), Autosomal recessive
ETFA	99.88 %	608053	Glutaric acidemia IIA, 231680 (3), Autosomal recessive
ETFB	100 %	130410	Glutaric acidemia IIB, 231680 (3), Autosomal recessive
FAH	99.98 %	613871	Tyrosinemia, type I, 276700 (3), Autosomal recessive
FAM111B	99.98 %	615584	Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis, 615704 (3), Autosomal dominant
FARSA	100 %	602918	?Rajab interstitial lung disease with brain calcifications 2, 619013 (3), Autosomal recessive
FARSB	99.64 %	609690	Rajab interstitial lung disease with brain calcifications 1, 613658 (3), Autosomal recessive
FBP1	100 %	611570	Fructose-1,6-bisphosphatase deficiency, 229700 (3), Autosomal recessive
FOCAD	99.62 %	614606	Liver disease, severe congenital, 619991 (3), Autosomal recessive
FTH1	22.62 %	134770	?Hemochromatosis, type 5, 615517 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
FTL	99.99 %	134790	Hyperferritinemia-cataract syndrome, 600886 (3), Autosomal dominant; L-ferritin deficiency, dominant and recessive, 615604 (3), Autosomal recessive, Autosomal dominant; Neurodegeneration with brain iron accumulation 3, 606159 (3), Autosomal dominant
G6PC1	99.93 %	613742	Glycogen storage disease Ia, 232200 (3), Autosomal recessive
GAA	100 %	606800	Glycogen storage disease II, 232300 (3), Autosomal recessive
GALE	99.9 %	606953	Galactose epimerase deficiency, 230350 (3), Autosomal recessive
GALK1	100 %	604313	Galactokinase deficiency with cataracts, 230200 (3), Autosomal recessive
GALM	100 %	137030	Galactosemia IV, 618881 (3), Autosomal recessive
GALT	100 %	606999	Galactosemia, 230400 (3), Autosomal recessive
GANAB	99.97 %	104160	Polycystic kidney disease 3, 600666 (3), Autosomal dominant
GBA	96.92 %	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
GBE1	99.73 %	607839	Glycogen storage disease IV, 232500 (3), Autosomal recessive; Polyglucosan body disease, adult form, 263570 (3), Autosomal recessive
GFM1	99.95 %	606639	Combined oxidative phosphorylation deficiency 1, 609060 (3), Autosomal recessive
GIMAP5	100 %	608086	Portal hypertension, noncirrhotic, 2, 619463 (3), Autosomal recessive
GLIS3	99.99 %	610192	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199 (3), Autosomal recessive
GNAS	100 %	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
GPD1	99.85 %	138420	Hypertriglyceridemia, transient infantile, 614480 (3), Autosomal recessive
GUSB	95.07 %	611499	Mucopolysaccharidosis VII, 253220 (3), Autosomal recessive
GYS1	99.98 %	138570	Glycogen storage disease 0, muscle, 611556 (3), Autosomal recessive
GYS2	99.86 %	138571	Glycogen storage disease 0, liver, 240600 (3), Autosomal recessive
HADHA	99.98 %	600890	HELLP syndrome, maternal, of pregnancy, 609016 (3), Autosomal recessive; LCHAD deficiency, 609016 (3), Autosomal recessive; Mitochondrial trifunctional protein deficiency 1, 609015 (3), Autosomal recessive; Fatty liver, acute, of pregnancy, 609016 (3), Autosomal recessive
HAMP	99.99 %	606464	Hemochromatosis, type 2B, 613313 (3), Autosomal recessive
HFE	100 %	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
HJV	99.99 %	608374	Hemochromatosis, type 2A, 602390 (3), Autosomal recessive
HLCS	99.97 %	609018	Holocarboxylase synthetase deficiency, 253270 (3), Autosomal recessive
HMOX1	99.95 %	141250	Heme oxygenase-1 deficiency, 614034 (3), Autosomal recessive; {Pulmonary disease, chronic obstructive, susceptibility to}, 606963 (3)

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
HNF1A	100 %	142410	Hepatic adenoma, somatic, 142330 (3); Diabetes mellitus, insulin-dependent, 20, 612520 (3); {Diabetes mellitus, noninsulin-dependent, 2}, 125853 (3), Autosomal dominant; MODY, type III, 600496 (3), Autosomal dominant; {Diabetes mellitus, insulin-dependent}, 222100 (3), Autosomal recessive; Renal cell carcinoma, 144700 (3)
HNF1B	100 %	189907	Type 2 diabetes mellitus, 125853 (3), Autosomal dominant; Renal cysts and diabetes syndrome, 137920 (3), Autosomal dominant; {Renal cell carcinoma}, 144700 (3)
HPS1	100 %	604982	Hermansky-Pudlak syndrome 1, 203300 (3), Autosomal recessive
HSD17B4	99.71 %	601860	D-bifunctional protein deficiency, 261515 (3), Autosomal recessive; Perrault syndrome 1, 233400 (3), Autosomal recessive
HSD3B7	100 %	607764	Bile acid synthesis defect, congenital, 1, 607765 (3), Autosomal recessive
IARS1	99.89 %	600709	Growth retardation, impaired intellectual development, hypotonia, and hepatopathy, 617093 (3), Autosomal recessive
IFT122	99.98 %	606045	Cranioectodermal dysplasia 1, 218330 (3), Autosomal recessive
IFT140	100 %	614620	Short-rib thoracic dysplasia 9 with or without polydactyly, 266920 (3), Autosomal recessive; Retinitis pigmentosa 80, 617781 (3), Autosomal recessive
IFT172	99.98 %	607386	Retinitis pigmentosa 71, 616394 (3), Autosomal recessive; Bardet-Biedl syndrome 20, 619471 (3), Autosomal recessive; Short-rib thoracic dysplasia 10 with or without polydactyly, 615630 (3), Autosomal recessive
INVS	99.94 %	243305	Nephronophthisis 2, infantile, 602088 (3), Autosomal recessive
ITCH	95.57 %	606409	Autoimmune disease, multisystem, with facial dysmorphism, 613385 (3), Autosomal recessive
IVD	100 %	607036	Isovaleric acidemia, 243500 (3), Autosomal recessive
JAG1	100 %	601920	?Deafness, congenital heart defects, and posterior embryotoxon, 617992 (3), Autosomal dominant; Charcot-Marie-Tooth disease, axonal, type 2HH, 619574 (3), Autosomal dominant; Alagille syndrome 1, 118450 (3), Autosomal dominant; Tetralogy of Fallot, 187500 (3), Autosomal dominant
KCNN3	99.97 %	602983	Zimmermann-Laband syndrome 3, 618658 (3), Autosomal dominant
KIF12	99.99 %	611278	Cholestasis, progressive familial intrahepatic, 8, 619662 (3), Autosomal recessive
KRT18	47.42 %	148070	Cirrhosis, cryptogenic, 215600 (3), Autosomal recessive; {Cirrhosis, noncryptogenic, susceptibility to}, 215600 (3), Autosomal recessive
KRT8	71.6 %	148060	*No OMIM phenotypes*
LAMP2	98.95 %	309060	Danon disease, 300257 (3), X-linked dominant
LARS1	99.87 %	151350	?Infantile liver failure syndrome 1, 615438 (3), Autosomal recessive
LDHA	99.94 %	150000	Glycogen storage disease XI, 612933 (3), Autosomal recessive
LIPA	99.96 %	613497	Wolman disease, 620151 (3), Autosomal recessive; Cholesteryl ester storage disease, 278000 (3), Autosomal recessive
LRP5	99.95 %	603506	Osteopetrosis, autosomal dominant 1, 607634 (3), Autosomal dominant; [Bone mineral density variability 1], 601884 (3), Autosomal dominant; Polycystic liver disease 4 with or without kidney cysts, 617875 (3), Autosomal dominant; Endosteal hyperostosis, 144750 (3), Autosomal dominant; Osteoporosis-pseudoglioma syndrome, 259770 (3), Autosomal recessive; Exudative vitreoretinopathy 4, 601813 (3), Autosomal recessive, Autosomal dominant
LRPPRC	99.8 %	607544	Mitochondrial complex IV deficiency, nuclear type 5, (French-Canadian), 220111 (3), Autosomal recessive
LSR	99.98 %	616582	*No OMIM phenotypes*

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MARS1	99.97 %	156560	Spastic paraplegia 70, autosomal recessive, 620323 (3), Autosomal recessive; Interstitial lung and liver disease, 615486 (3), Autosomal recessive; ?Trichothiodystrophy 9, nonphotosensitive, 619692 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal, type 2U, 616280 (3), Autosomal dominant
MCEE	99.9 %	608419	Methylmalonyl-CoA epimerase deficiency, 251120 (3), Autosomal recessive
MICOS13	99.95 %	616658	Combined oxidative phosphorylation deficiency 37, 618329 (3), Autosomal recessive
MKS1	99.92 %	609883	Bardet-Biedl syndrome 13, 615990 (3), Autosomal recessive; Meckel syndrome 1, 249000 (3), Autosomal recessive; Joubert syndrome 28, 617121 (3), Autosomal recessive
MMUT	99.68 %	609058	Methylmalonic aciduria, mut(0) type, 251000 (3), Autosomal recessive
MPI	99.95 %	154550	Congenital disorder of glycosylation, type Ib, 602579 (3), Autosomal recessive
MPV17	99.98 %	137960	Charcot-Marie-Tooth disease, axonal, type 2EE, 618400 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810 (3), Autosomal recessive
MRPS23	100 %	611985	?Combined oxidative phosphorylation deficiency 46, 618952 (3), Autosomal recessive
MVK	99.97 %	251170	Hyper-IgD syndrome, 260920 (3), Autosomal recessive; Porokeratosis 3, multiple types, 175900 (3), Autosomal dominant; Mevalonic aciduria, 610377 (3), Autosomal recessive
MYO5B	100 %	606540	Diarrhea 2, with microvillus atrophy, with or without cholestasis, 251850 (3), Autosomal recessive; Cholestasis, progressive familial intrahepatic, 10, 619868 (3), Autosomal recessive
NBAS	99.86 %	608025	Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800 (3), Autosomal recessive; Infantile liver failure syndrome 2, 616483 (3), Autosomal recessive
NCOA6	99.99 %	605299	*No OMIM phenotypes*
NEK8	99.99 %	609799	Renal-hepatic-pancreatic dysplasia 2, 615415 (3), Autosomal recessive; ?Nephronophthisis 9, 613824 (3)
NEU1	99.98 %	608272	Sialidosis, type II, 256550 (3), Autosomal recessive; Sialidosis, type I, 256550 (3), Autosomal recessive
NGLY1	99.93 %	610661	Congenital disorder of deglycosylation 1, 615273 (3), Autosomal recessive
NHLRC1	100 %	608072	Epilepsy, progressive myoclonic 2B (Lafora), 254780 (3), Autosomal recessive
NOTCH2	99.03 %	600275	Alagille syndrome 2, 610205 (3), Autosomal dominant; Hajdu-Cheney syndrome, 102500 (3), Autosomal dominant
NPC1	99.99 %	607623	Niemann-Pick disease, type C1, 257220 (3), Autosomal recessive; Niemann-Pick disease, type D, 257220 (3), Autosomal recessive
NPC2	100 %	601015	Niemann-pick disease, type C2, 607625 (3), Autosomal recessive
NPHP1	99.05 %	607100	Joubert syndrome 4, 609583 (3), Autosomal recessive; Nephronophthisis 1, juvenile, 256100 (3), Autosomal recessive; Senior-Loken syndrome-1, 266900 (3), Autosomal recessive
NPHP3	99.89 %	608002	Nephronophthisis 3, 604387 (3), Autosomal recessive; Renal-hepatic-pancreatic dysplasia 1, 208540 (3), Autosomal recessive; Meckel syndrome 7, 267010 (3), Autosomal recessive
NPHP4	99.98 %	607215	Senior-Loken syndrome 4, 606996 (3), Autosomal recessive; Nephronophthisis 4, 606966 (3), Autosomal recessive
NR1H4	99.49 %	603826	Cholestasis, progressive familial intrahepatic, 5, 617049 (3), Autosomal recessive
OTC	99.42 %	300461	Ornithine transcarbamylase deficiency, 311250 (3), X-linked
OXCT1	99.82 %	601424	Succinyl CoA:3-oxoacid CoA transferase deficiency, 245050 (3), Autosomal recessive
PC	99.99 %	608786	Pyruvate carboxylase deficiency, 266150 (3), Autosomal recessive
PCCA	99.9 %	232000	Propionicacidemia, 606054 (3), Autosomal recessive

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PCCB	99.97 %	232050	Propionicacidemia, 606054 (3), Autosomal recessive
PCK1	100 %	614168	Phosphoenolpyruvate carboxykinase deficiency, cytosolic, 261680 (3), Autosomal recessive
PEX1	98.8 %	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
PEX10	100 %	602859	Peroxisome biogenesis disorder 6A (Zellweger), 614870 (3), Autosomal recessive; Peroxisome biogenesis disorder 6B, 614871 (3), Autosomal recessive
PEX11B	99.62 %	603867	Peroxisome biogenesis disorder 14B, 614920 (3), Autosomal recessive
PEX12	100 %	601758	Peroxisome biogenesis disorder 3B, 266510 (3), Autosomal recessive; Peroxisome biogenesis disorder 3A (Zellweger), 614859 (3), Autosomal recessive
PEX13	99.36 %	601789	Peroxisome biogenesis disorder 11A (Zellweger), 614883 (3), Autosomal recessive; Peroxisome biogenesis disorder 11B, 614885 (3), Autosomal recessive
PEX14	100 %	601791	Peroxisome biogenesis disorder 13A (Zellweger), 614887 (3), Autosomal recessive
PEX16	99.94 %	603360	Peroxisome biogenesis disorder 8B, 614877 (3), Autosomal recessive; Peroxisome biogenesis disorder 8A (Zellweger), 614876 (3), Autosomal recessive
PEX19	99.25 %	600279	Peroxisome biogenesis disorder 12A (Zellweger), 614886 (3), Autosomal recessive
PEX2	100 %	170993	Peroxisome biogenesis disorder 5A (Zellweger), 614866 (3), Autosomal recessive; Peroxisome biogenesis disorder 5B, 614867 (3), Autosomal recessive
PEX26	100 %	608666	Peroxisome biogenesis disorder 7B, 614873 (3), Autosomal recessive; Peroxisome biogenesis disorder 7A (Zellweger), 614872 (3), Autosomal recessive
PEX3	99.85 %	603164	Peroxisome biogenesis disorder 10A (Zellweger), 614882 (3), Autosomal recessive; ?Peroxisome biogenesis disorder 10B, 617370 (3), Autosomal recessive
PEX5	99.89 %	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
PEX6	99.99 %	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
PEX7	99.72 %	601757	Rhizomelic chondrodysplasia punctata, type 1, 215100 (3), Autosomal recessive; Peroxisome biogenesis disorder 9B, 614879 (3), Autosomal recessive
PFKM	99.57 %	610681	Glycogen storage disease VII, 232800 (3), Autosomal recessive
PGAM2	100 %	612931	Glycogen storage disease X, 261670 (3), Autosomal recessive
PGK1	99.93 %	311800	Phosphoglycerate kinase 1 deficiency, 300653 (3), X-linked recessive
PGM1	96.77 %	171900	Congenital disorder of glycosylation, type It, 614921 (3), Autosomal recessive
PHKA1	99.84 %	311870	Muscle glycogenosis, 300559 (3), X-linked recessive
PHKA2	99.92 %	300798	Glycogen storage disease, type IXa2, 306000 (3), X-linked recessive; Glycogen storage disease, type IXa1, 306000 (3), X-linked recessive
PHKB	99.69 %	172490	Phosphorylase kinase deficiency of liver and muscle, autosomal recessive, 261750 (3), Autosomal recessive
PHKG2	99.86 %	172471	Glycogen storage disease IXc, 613027 (3), Autosomal recessive
PKD1	99.98 %	601313	Polycystic kidney disease 1, 173900 (3), Autosomal dominant
PKD2	99.91 %	173910	Polycystic kidney disease 2, 613095 (3), Autosomal dominant
PKHD1	99.95 %	606702	Polycystic kidney disease 4, with or without hepatic disease, 263200 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
PLEC	100 %	601282	?Epidermolysis bullosa simplex 5D, generalized intermediate, autosomal recessive, 616487 (3), Autosomal recessive; Epidermolysis bullosa simplex 5B, with muscular dystrophy, 226670 (3), Autosomal recessive; Epidermolysis bullosa simplex 5C, with pyloric atresia, 612138 (3), Autosomal recessive; Epidermolysis bullosa simplex 5A, Ogna type, 131950 (3), Autosomal dominant; Muscular dystrophy, limb-girdle, autosomal recessive 17, 613723 (3), Autosomal recessive
PMM2	99.93 %	601785	Congenital disorder of glycosylation, type Ia, 212065 (3), Autosomal recessive
PNPLA3	100 %	609567	*No OMIM phenotypes*
POLG	100 %	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
PPM1F	100 %	619309	*No OMIM phenotypes*
PRKAG2	99.96 %	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
PRKCSH	99.99 %	177060	Polycystic liver disease 1, 174050 (3), Autosomal dominant
PROC	99.98 %	612283	Thrombophilia 3 due to protein C deficiency, autosomal dominant, 176860 (3), Autosomal dominant; Thrombophilia 3 due to protein C deficiency, autosomal recessive, 612304 (3), Autosomal recessive
PYGL	99.99 %	613741	Glycogen storage disease VI, 232700 (3), Autosomal recessive
PYGM	99.96 %	608455	McArdle disease, 232600 (3), Autosomal recessive
QRSL1	99.97 %	617209	Combined oxidative phosphorylation deficiency 40, 618835 (3), Autosomal recessive
RBCK1	100 %	610924	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895 (3), Autosomal recessive
RINT1	99.99 %	610089	Infantile liver failure syndrome 3, 618641 (3), Autosomal recessive
RPGRIP1L	96.35 %	610937	Joubert syndrome 7, 611560 (3), Autosomal recessive; Meckel syndrome 5, 611561 (3), Autosomal recessive; ?COACH syndrome 3, 619113 (3), Autosomal recessive
SBDS	99.93 %	607444	{Aplastic anemia, susceptibility to}, 609135 (3); Shwachman-Diamond syndrome 1, 260400 (3), Autosomal recessive
SCO1	99.98 %	603644	Mitochondrial complex IV deficiency, nuclear type 4, 619048 (3), Autosomal recessive
SCYL1	100 %	607982	Spinocerebellar ataxia, autosomal recessive 21, 616719 (3), Autosomal recessive
SEC23B	99.93 %	610512	?Cowden syndrome 7, 616858 (3), Autosomal dominant; Dyserythropoietic anemia, congenital, type II, 224100 (3), Autosomal recessive
SEC61B	99.99 %	609214	*No OMIM phenotypes*
SEC63	99.84 %	608648	Polycystic liver disease 2, 617004 (3), Autosomal dominant
SEMA7A	99.9 %	607961	?Cholestasis, progressive familial intrahepatic, 11, 619874 (3), Autosomal recessive; [Blood group, John-Milton-Hagen system], 614745 (3)
SERAC1	99.9 %	614725	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739 (3), Autosomal recessive
SERPINA1	100 %	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
SKIV2L	99.98 %	600478	Trichohepatoenteric syndrome 2, 614602 (3), Autosomal recessive
SLC10A1	99.99 %	182396	Hypercholanemia, familial 2, 619256 (3), Autosomal recessive
SLC10A2	99.99 %	601295	?Bile acid malabsorption, primary, 1, 613291 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
SLC11A2	99.68 %	600523	Anemia, hypochromic microcytic, with iron overload 1, 206100 (3), Autosomal recessive
SLC16A1	99.26 %	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
SLC25A13	99.67 %	603859	Citrullinemia, type II, neonatal-onset, 605814 (3), Autosomal recessive; Citrullinemia, adult-onset type II, 603471 (3), Autosomal recessive
SLC25A20	100 %	613698	Carnitine-acylcarnitine translocase deficiency, 212138 (3), Autosomal recessive
SLC25A38	99.98 %	610819	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950 (3), Autosomal recessive
SLC2A2	99.96 %	138160	Fanconi-Bickel syndrome, 227810 (3), Autosomal recessive; {Diabetes mellitus, noninsulin-dependent}, 125853 (3), Autosomal dominant
SLC30A10	99.99 %	611146	Hypermanganesemia with dystonia 1, 613280 (3), Autosomal recessive
SLC37A4	99.9 %	602671	Glycogen storage disease Ib, 232220 (3), Autosomal recessive; Congenital disorder of glycosylation, type IIw, 619525 (3), Autosomal dominant; Glycogen storage disease Ic, 232240 (3), Autosomal recessive
SLC40A1	99.7 %	604653	Hemochromatosis, type 4, 606069 (3), Autosomal dominant
SLC51A	100 %	612084	?Cholestasis, progressive familial intrahepatic, 6, 619484 (3), Autosomal recessive
SLCO1B1	98.69 %	604843	Hyperbilirubinemia, Rotor type, digenic, 237450 (3), Digenic recessive
SLCO1B3	99.85 %	605495	Hyperbilirubinemia, Rotor type, digenic, 237450 (3), Digenic recessive
SMPD1	100 %	607608	Niemann-Pick disease, type B, 607616 (3), Autosomal recessive; Niemann-Pick disease, type A, 257200 (3), Autosomal recessive
SP110	99.99 %	604457	{Mycobacterium tuberculosis, susceptibility to}, 607948 (3); Hepatic venoocclusive disease with immunodeficiency, 235550 (3), Autosomal recessive
STAB1	99.98 %	608560	*No OMIM phenotypes*
STEAP3	100 %	609671	?Anemia, hypochromic microcytic, with iron overload 2, 615234 (3), Autosomal dominant
STN1	99.88 %	613128	Cerebroretinal microangiopathy with calcifications and cysts 2, 617341 (3), Autosomal recessive
STT3B	99.96 %	608605	Congenital disorder of glycosylation, type Ix, 615597 (3), Autosomal recessive
SUCLG1	99.64 %	611224	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400 (3), Autosomal recessive
TALDO1	100 %	602063	Transaldolase deficiency, 606003 (3), Autosomal recessive
TANGO2	99.85 %	616830	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878 (3), Autosomal recessive
TF	99.96 %	190000	Atransferrinemia, 209300 (3), Autosomal recessive
TFR2	99.97 %	604720	Hemochromatosis, type 3, 604250 (3), Autosomal recessive
TJP2	99.99 %	607709	Hypercholanemia, familial 1, 607748 (3), Autosomal recessive; Cholestasis, progressive familial intrahepatic 4, 615878 (3), Autosomal recessive
TM6SF2	99.99 %	606563	*No OMIM phenotypes*
TMEM216	99.98 %	613277	Joubert syndrome 2, 608091 (3), Autosomal recessive; Meckel syndrome 2, 603194 (3), Autosomal recessive
TMEM67	99.69 %	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
TMPRSS6	100 %	609862	Iron-refractory iron deficiency anemia, 206200 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
TRMU	100 %	610230	{Deafness, mitochondrial, modifier of}, 580000 (3), Mitochondrial; Liver failure, transient infantile, 613070 (3), Autosomal recessive
TTC37	99.82 %	614589	Trichohepatoenteric syndrome 1, 222470 (3), Autosomal recessive
TULP3	99.91 %	604730	Hepatorenocardiac degenerative fibrosis, 619902 (3), Autosomal recessive
TWNK	100 %	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
UGT1A1	99.98 %	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, Autosomal dominant; Crigler-Najjar syndrome, type II, 606785 (3), Autosomal recessive; [Gilbert syndrome], 143500 (3), Autosomal recessive
UNC45A	100 %	611219	Osteootohepatoenteric syndrome, 619377 (3), Autosomal recessive
UQCRC2	99.49 %	191329	Mitochondrial complex III deficiency, nuclear type 5, 615160 (3), Autosomal recessive
UROD	99.32 %	613521	Porphyria, hepatoerythropoietic, 176100 (3), Autosomal recessive, Autosomal dominant; Porphyria cutanea tarda, 176100 (3), Autosomal recessive, Autosomal dominant
UROS	100 %	606938	Porphyria, congenital erythropoietic, 263700 (3), Autosomal recessive
USP24	96.06 %	610569	*No OMIM phenotypes*
USP53	99.92 %	617431	Cholestasis, progressive familial intrahepatic, 7, with or without hearing loss, 619658 (3), Autosomal recessive
UTP4	99.91 %	607456	*No OMIM phenotypes*
VIPAS39	99.87 %	613401	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404 (3), Autosomal recessive
VPS33B	99.95 %	608552	Keratoderma-ichthyosis-deafness syndrome, autosomal recessive, 620009 (3), Autosomal recessive; Cholestasis, progressive familial intrahepatic, 12, 620010 (3), Autosomal recessive; Arthrogryposis, renal dysfunction, and cholestasis 1, 208085 (3), Autosomal recessive
VPS50	98.35 %	616465	Neurodevelopmental disorder with microcephaly, seizures, and neonatal cholestasis, 619685 (3), Autosomal recessive
WDR83OS	100 %	618474	*No OMIM phenotypes*
YARS1	99.29 %	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
ZFYVE19	99.99 %	619635	Cholestasis, progressive familial intrahepatic, 9, 619849 (3), Autosomal recessive

Explanation

OMIM release used for OMIM disease identifiers and descriptions: **2023-07-31**

Gene symbols used are according to the HGNC guidelines (corresponding to Ensembl database release 105).

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

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.

* The column '% at least 20 x covered' shows the percentage of the coding sequence (+/-20 nucleotides of the flanking introns) of that gene that is on average at least 20 x covered. This according to the experience with exome sequencing in our laboratory and based on the current method.