LIVER DISORDERS GENE PANEL DG 3.1.0 (126 genes)

Releasedate: 23-03-2021

Gene	*Agilent V5 covered >10x*	*Agilent V5 covered >20x*	*TWIST covered >10x*	*TWIST covered >20x*	*Associated Phenotype description and OMIM disease ID*
ABCB11	100	99,7	100	100	Cholestasis, progressive familial intrahepatic 2, 601847 Cholestasis, benign recurrent intrahepatic, 2, 605479
ABCB4	99,9	99,6	100	100	Gallbladder disease 1, 600803 Cholestasis, intrahepatic, of pregnancy, 3, 614972 Cholestasis, progressive familial intrahepatic 3, 602347
ABCC2	100	99,9	100	100	Dubin-Johnson syndrome, 237500
ABCD3	99,8	97,7	100	100	?Bile acid synthesis defect, congenital, 5, 616278
ACOX2	100	99,2	100	100	Bile acid synthesis defect, congenital, 6, 617308
ACTA2	100	99	100	100	Aortic aneurysm, familial thoracic 6, 611788 Multisystemic smooth muscle dysfunction syndrome, 613834 Moyamoya disease 5, 614042
ACTG2	99,9	98,2	100	100	Visceral myopathy, 155310
ADK	84,1	81	84,5	84,5	Hypermethioninemia due to adenosine kinase deficiency, 614300
AHCY	100	99,2	100	100	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752
AKR1D1	100	99,4	100	100	Bile acid synthesis defect, congenital, 2, 235555
ALDOB	99,4	96,6	100	100	Fructose intolerance, hereditary, 229600
ALG8	97,2	95,6	96,6	96,6	Congenital disorder of glycosylation, type Ih, 608104 Polycystic liver disease 3 with or without kidney cysts, 617874
AMACR	100	100	100	100	Bile acid synthesis defect, congenital, 4, 214950 Alpha-methylacyl-CoA racemase deficiency, 614307
ANKS6	93,8	89,5	97,9	95,8	Nephronophthisis 16, 615382
AP1S1	99,9	99,5	100	100	MEDNIK syndrome, 609313
ATP7B	99,9	99,2	100	100	Wilson disease, 277900
ATP8B1	96,5	94	100	100	Cholestasis, intrahepatic, of pregnancy, 1, 147480 Cholestasis, progressive familial intrahepatic 1, 211600 Cholestasis, benign recurrent intrahepatic, 243300
BAAT	99,8	98,4	100	100	Hypercholanemia, familial, 607748

BCS1L	100	100	100	100	GRACILE syndrome, 603358 Bjornstad syndrome, 262000 Mitochondrial complex III deficiency, nuclear type 1, 124000
BLVRA	100	99,4	100	100	Hyperbiliverdinemia, 614156
CC2D2A	98,5	96,5	97,1	97,1	Meckel syndrome 6, 612284 Joubert syndrome 9, 612285 COACH syndrome 2, 619111
CEP83	99,8	97,4	100	100	Nephronophthisis 18, 615862
CFC1	84,2	74,1	100	100	Heterotaxy, visceral, 2, autosomal, 605376
CFTR	99,6	97,9	100	100	{Bronchiectasis with or without elevated sweat chloride 1, modifier of}, 211400 Cystic fibrosis, 219700 Congenital bilateral absence of vas deferens, 277180 {Pancreatitis, hereditary}, 167800 Sweat chloride elevation without CF, 0 {Hypertrypsinemia, neonatal}, 0
CHD8	100	99,9	100	100	{Autism, susceptibility to, 18}, 615032
CHRM3	100	100	100	100	Prune belly syndrome, 100100
CHRNA3	100	99,4	100	100	{Lung cancer susceptibility 2}, 612052 Bladder dysfunction, autonomic, with impaired pupillary reflex and secondary CAKUT, 191800
CLDN1	100	100	100	100	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis, 607626
CLMP	100	99,6	100	100	Congenital short bowel syndrome, 615237
COG7	100	100	100	100	Congenital disorder of glycosylation, type IIe, 608779
CYP27A1	98,9	96,7	100	100	Cerebrotendinous xanthomatosis, 213700
CYP7B1	98	92,8	100	100	Spastic paraplegia 5A, autosomal recessive, 270800 Bile acid synthesis defect, congenital, 3, 613812
DCDC2	100	99,9	100	100	Sclerosing cholangitis, neonatal, 617394 Nephronophthisis 19, 616217 ?Deafness, autosomal recessive 66, 610212
DGUOK	100	99,4	100	100	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, 617070 Portal hypertension, noncirrhotic, 617068 Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880
DHCR7	100	100	100	100	Smith-Lemli-Opitz syndrome, 270400
DKC1	99,8	98,7	100	99,7	Dyskeratosis congenita, X-linked, 305000
DNAJB11	100	99,5	100	100	Polycystic kidney disease 6 with or without polycystic liver disease, 618061
EDNRB	98	93,8	100	100	{Hirschsprung disease, susceptibility to, 2}, 600155 Waardenburg syndrome, type 4A, 277580 ABCD syndrome, 600501
EPHX1	99,9	98,8	100	100	?Hypercholanemia, familial, 607748

ETFDH	100	99,8	100	100	Glutaric acidemia IIC, 231680
FAH	100	100	100	100	Tyrosinemia, type I, 276700
FECH	100	100	100	100	Protoporphyria, erythropoietic, 1, 177000
FH	92,1	88,3	100	100	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800
FLNA	100	99,9	100	100	Otopalatodigital syndrome, type I, 311300 Congenital short bowel syndrome, 300048 Otopalatodigital syndrome, type II, 304120 Intestinal pseudoobstruction, neuronal, 300048 Melnick-Needles syndrome, 309350 Cardiac valvular dysplasia, X-linked, 314400 ?FG syndrome 2, 300321 Heterotopia, periventricular, 1, 300049 Terminal osseous dysplasia, 300244 Frontometaphyseal dysplasia 1, 305620
GALT	100	99,7	100	100	Galactosemia, 230400
GANAB	99,9	99	100	100	Polycystic kidney disease 3, 600666
GBA	100	100	100	100	Gaucher disease, type III, 231000 {Parkinson disease, late-onset, susceptibility to}, 168600 Gaucher disease, type IIIC, 231005 Gaucher disease, type I, 230800 Gaucher disease, perinatal lethal, 608013 Gaucher disease, type II, 230900 {Lewy body dementia, susceptibility to}, 127750
GBE1	100	99,6	100	100	Polyglucosan body disease, adult form, 263570 Glycogen storage disease IV, 232500
GDNF	100	100	100	100	{Pheochromocytoma, modifier of}, 171300 {Hirschsprung disease, susceptibility to, 3}, 613711 Central hypoventilation syndrome, 209880
GFM1	99,9	99,4	100	100	Combined oxidative phosphorylation deficiency 1, 609060
GLI3	98,5	98	100	100	Polydactyly, postaxial, types A1 and B, 174200 Greig cephalopolysyndactyly syndrome, 175700 Polydactyly, preaxial, type IV, 174700 Pallister-Hall syndrome, 146510
GLIS3	98,6	98,2	100	100	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199
HADHA	97,2	91,6	100	100	LCHAD deficiency, 609016 HELLP syndrome, maternal, of pregnancy, 609016

					Mitochondrial trifunctional protein deficiency, 609015 Fatty liver, acute, of pregnancy, 609016
HAMP	100	100	100	100	Hemochromatosis, type 2B, 613313
HFE	100	99,7	100	100	{Porphyria variegata, susceptibility to}, 176200 {Microvascular complications of diabetes 7}, 612635 {Porphyria cutanea tarda, susceptibility to}, 176100 [Transferrin serum level QTL2], 614193 {Alzheimer disease, susceptibility to}, 104300 Hemochromatosis, 235200
HNF1B	99,3	96,1	100	100	Renal cysts and diabetes syndrome, 137920 {Renal cell carcinoma}, 144700 Type 2 diabetes mellitus, 125853
HSD17B4	95,4	93,1	96,6	96,6	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
HSD3B7	99,1	95,5	100	100	Bile acid synthesis defect, congenital, 1, 607765
IARS1	100	99,6	100	100	Growth retardation, impaired intellectual development, hypotonia, and hepatopathy, 617093
IFT140	99,8	98,8	100	100	Retinitis pigmentosa 80, 617781 Short-rib thoracic dysplasia 9 with or without polydactyly, 266920
IFT172	99,9	99,1	100	100	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT43	100	100	100	100	?Cranioectodermal dysplasia 3, 614099 Short-rib thoracic dysplasia 18 with polydactyly, 617866 ?Retinitis pigmentosa 81, 617871
INSR	97,8	94,7	99,9	99,2	Hyperinsulinemic hypoglycemia, familial, 5, 609968 Rabson-Mendenhall syndrome, 262190 Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549 Leprechaunism, 246200
INVS	100	100	100	100	Nephronophthisis 2, infantile, 602088
JAG1	97,7	96,8	100	100	?Deafness, congenital heart defects, and posterior embryotoxon, 617992 Alagille syndrome 1, 118450 Tetralogy of Fallot, 187500
LARS1	99,8	98,4	100	100	?Infantile liver failure syndrome 1, 615438
LRP5	98,5	98,1	100	99,7	van Buchem disease, type 2, 607636 Exudative vitreoretinopathy 4, 601813 Hyperostosis, endosteal, 144750 Osteosclerosis, 144750 Polycystic liver disease 4 with or without kidney cysts, 617875 Osteoporosis-pseudoglioma syndrome, 259770

					Osteopetrosis, autosomal dominant 1, 607634 {Osteoporosis}, 166710 [Bone mineral density variability 1], 601884
MARS1	99,7	97,4	100	100	Charcot-Marie-Tooth disease, axonal, type 2U, 616280 Interstitial lung and liver disease, 615486
MPV17	100	97,2	100	100	Charcot-Marie-Tooth disease, axonal, type 2EE, 618400 Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810
MTM1	99	93,3	100	100	Myotubular myopathy, X-linked, 310400
MYO5B	99,1	96,2	100	100	Microvillus inclusion disease, 251850
NBAS	100	99,6	100	100	Infantile liver failure syndrome 2, 616483 Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800
NHP2	100	100	100	100	Dyskeratosis congenita, autosomal recessive 2, 613987
NOP10	100	99,8	100	100	Dyskeratosis congenita, autosomal recessive 1, 224230
NOTCH2	100	99,5	100	100	Hajdu-Cheney syndrome, 102500 Alagille syndrome 2, 610205
NPC1	99,6	98,7	100	100	Niemann-Pick disease, type D, 257220 Niemann-Pick disease, type C1, 257220
NPC2	100	99,6	100	100	Niemann-pick disease, type C2, 607625
NPHP3	99,7	98,4	100	100	Meckel syndrome 7, 267010 Renal-hepatic-pancreatic dysplasia 1, 208540 Nephronophthisis 3, 604387
NR1H4	99,8	98,5	100	100	Cholestasis, progressive familial intrahepatic, 5, 617049
PEX1	99,9	99,4	100	100	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 Peroxisome biogenesis disorder 1A (Zellweger), 214100
PEX10	96,8	89,7	100	99,9	Peroxisome biogenesis disorder 6B, 614871 Peroxisome biogenesis disorder 6A (Zellweger), 614870
PEX12	100	100	100	100	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B, 266510
PEX13	100	100	100	100	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885
PEX14	96,7	90,8	100	100	Peroxisome biogenesis disorder 13A (Zellweger), 614887
PEX16	97,9	94,2	100	100	Peroxisome biogenesis disorder 8A (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877
PEX19	99,9	98,5	100	100	Peroxisome biogenesis disorder 12A (Zellweger), 614886
PEX2	100	100	100	100	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867

PEX26	100	100	100	100	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873
PEX3	100	99,3	100	100	Peroxisome biogenesis disorder 10A (Zellweger), 614882 ?Peroxisome biogenesis disorder 10B, 617370
PEX5	99,9	99	100	100	Peroxisome biogenesis disorder 2B, 202370 Rhizomelic chondrodysplasia punctata, type 5, 616716 Peroxisome biogenesis disorder 2A (Zellweger), 214110
PEX6	94,5	86,7	100	100	Peroxisome biogenesis disorder 4B, 614863 Heimler syndrome 2, 616617 Peroxisome biogenesis disorder 4A (Zellweger), 614862
PEX7	87,8	80,7	91,3	91,3	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100
PKD1	39,2	30	99,2	98,9	Polycystic kidney disease 1, 173900
PKD2	95,5	91,1	99,3	97,7	Polycystic kidney disease 2, 613095
PKHD1	100	99,6	100	100	Polycystic kidney disease 4, with or without hepatic disease, 263200
POLG	100	99,3	100	100	Progressive external ophthalmoplegia, autosomal dominant 1, 157640 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 Progressive external ophthalmoplegia, autosomal recessive 1, 258450
POMC	100	100	100	100	{Obesity, early-onset, susceptibility to}, 601665 Obesity, adrenal insufficiency, and red hair due to POMC deficiency, 609734
PRKCSH	99,8	95,4	100	100	Polycystic liver disease 1, 174050
RAD21	99,2	96,6	100	100	?Mungan syndrome, 611376 Cornelia de Lange syndrome 4, 614701
RFX6	100	99,6	100	100	Mitchell-Riley syndrome, 615710
RPGRIP1L	96,7	95,7	100	99,5	?COACH syndrome 3, 619113 Meckel syndrome 5, 611561 Joubert syndrome 7, 611560
SC5D	100	99,5	100	100	Lathosterolosis, 607330
SCO1	97,1	93,8	100	100	Mitochondrial complex IV deficiency, nuclear type 4, 619048
SEC61B	99,1	92,4	100	100	No OMIM disease ID
SEC63	91,2	83,3	100	100	Polycystic liver disease 2, 617004
SERPINA1	100	100	100	100	Hemorrhagic diathesis due to antithrombin Pittsburgh, 613490 Emphysema-cirrhosis, due to AAT deficiency, 613490 Emphysema due to AAT deficiency, 613490
SGO1	99,9	98,9	100	100	Chronic atrial and intestinal dysrhythmia, 616201

SLC25A13	100	99,7	100	100	Citrullinemia, adult-onset type II, 603471 Citrullinemia, type II, neonatal-onset, 605814
SLC40A1	100	99,5	100	100	Hemochromatosis, type 4, 606069
SMPD1	100	100	100	100	Niemann-Pick disease, type A, 257200 Niemann-Pick disease, type B, 607616
STN1	100	100	100	100	Cerebroretinal microangiopathy with calcifications and cysts 2, 617341
TALDO1	100	97,9	100	100	Transaldolase deficiency, 606003
TERC	NC	NC	NC	NC	{Aplastic anemia}, 614743 Dyskeratosis congenita, autosomal dominant 1, 127550 {Pulmonary fibrosis, idiopathic, susceptibility to}, 614743
TERT	96,2	94,5	100	100	{Melanoma, cutaneous malignant, 9}, 615134 {Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1}, 614742 {Leukemia, acute myeloid}, 601626 {Dyskeratosis congenita, autosomal recessive 4}, 613989 {Dyskeratosis congenita, autosomal dominant 2}, 613989
TFR2	99,1	97,8	100	100	Hemochromatosis, type 3, 604250
TJP2	92,8	92,5	98,8	98,8	Hypercholanemia, familial, 607748 Cholestasis, progressive familial intrahepatic 4, 615878
TMEM67	99,5	95	100	99,9	Meckel syndrome 3, 607361 COACH syndrome 1, 216360 ?RHYNS syndrome, 602152 Nephronophthisis 11, 613550 {Bardet-Biedl syndrome 14, modifier of}, 615991 Joubert syndrome 6, 610688
TRAF3IP1	99,6	97,6	100	100	Senior-Loken syndrome 9, 616629
TRMU	100	100	100	99,9	Liver failure, transient infantile, 613070 {Deafness, mitochondrial, modifier of}, 580000
TTC37	100	99,3	100	100	Trichohepatoenteric syndrome 1, 222470
TWNK	100	100	100	100	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286 Perrault syndrome 5, 616138
TYMP	100	97	100	100	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041
UBR1	99,9	99,1	98	98	Johanson-Blizzard syndrome, 243800
UGT1A1	100	100	100	100	[Gilbert syndrome], 143500 Hyperbilirubinemia, familial transient neonatal, 237900 Crigler-Najjar syndrome, type I, 218800

					Crigler-Najjar syndrome, type II, 606785 [Bilirubin, serum level of, QTL1], 601816
VIPAS39	100	100	100	100	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VPS33B	100	100	100	100	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085

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-protein coding genes.
non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : March 23rd , 2021.
This list is accurate for panel version DG 3.1.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors