LIVER DISORDERS GENE PANEL DG 3.2.0 (128 genes)

Releasedate: 16-09-2021

Gene	*Agilent V5 covered >10x*	*Agilent V5 covered >20x*	*TWIST covered >10x*	*TWIST covered >20x*	*Associated Phenotype Description and OMIM disease ID*
ABCB11	100	99,4	100	100	Cholestasis, benign recurrent intrahepatic, 2, 605479 Cholestasis, progressive familial intrahepatic 2, 601847
ABCB4	99,9	99,3	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	99,4	98	100	100	?Bile acid synthesis defect, congenital, 5, 616278
ACOX2	100	99	100	100	Bile acid synthesis defect, congenital, 6, 617308
ACTA2	100	98,9	100	100	Multisystemic smooth muscle dysfunction syndrome, 613834 Aortic aneurysm, familial thoracic 6, 611788 Moyamoya disease 5, 614042
ACTG2	99,7	97,4	100	100	Megacystis-microcolon-intestinal hypoperistalsis syndrome 5, 619431 Visceral myopathy 1, 155310
ADK	83,3	79,7	84,5	84,5	Hypermethioninemia due to adenosine kinase deficiency, 614300
AHCY	99,9	98,8	100	100	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752
AKR1D1	99,8	98,6	100	100	Bile acid synthesis defect, congenital, 2, 235555
ALDOB	98,8	95,7	100	100	Fructose intolerance, hereditary, 229600
ALG8	96,6	95,9	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	Alpha-methylacyl-CoA racemase deficiency, 614307 Bile acid synthesis defect, congenital, 4, 214950
ANKS6	94,2	89,7	97	95	Nephronophthisis 16, 615382
AP1S1	99,9	99,4	100	100	MEDNIK syndrome, 609313
ATP7B	99,9	99,2	100	100	Wilson disease, 277900
ATP8B1	96,6	93,5	100	100	Cholestasis, progressive familial intrahepatic 1, 211600 Cholestasis, intrahepatic, of pregnancy, 1, 147480 Cholestasis, benign recurrent intrahepatic, 243300

BAAT	99,5	97,5	100	100	Hypercholanemia, familial, 607748 Bile acid conjugation defect 1, 619232
BCS1L	100	100	100	100	GRACILE syndrome, 603358 Mitochondrial complex III deficiency, nuclear type 1, 124000 Bjornstad syndrome, 262000
BLVRA	99,8	97,8	100	100	Hyperbiliverdinemia, 614156
CC2D2A	98,3	96,6	97,1	97	COACH syndrome 2, 619111 Meckel syndrome 6, 612284 Joubert syndrome 9, 612285
CEP83	99	96,6	100	99,9	Nephronophthisis 18, 615862
CFC1	85	78	100	100	Heterotaxy, visceral, 2, autosomal, 605376
CFTR	99,5	97,9	100	100	Cystic fibrosis, 219700 Congenital bilateral absence of vas deferens, 277180 Sweat chloride elevation without CF,
CHD8	100	99,7	100	100	No OMIM disease ID
CHRM3	100	100	100	100	Prune belly syndrome, 100100
CHRNA3	100	99,2	100	100	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	99,5	100	100	Congenital short bowel syndrome, 615237
COG7	100	99,4	100	100	Congenital disorder of glycosylation, type IIe, 608779
CYP27A1	99,7	98,1	100	100	Cerebrotendinous xanthomatosis, 213700
CYP7B1	98,1	92,7	100	100	Spastic paraplegia 5A, autosomal recessive, 270800 Bile acid synthesis defect, congenital, 3, 613812
DCDC2	100	99,9	100	100	Nephronophthisis 19, 616217 ?Deafness, autosomal recessive 66, 610212 Sclerosing cholangitis, neonatal, 617394
DGUOK	99,9	98,8	100	100	Portal hypertension, noncirrhotic, 1, 617068 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, 617070 Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880
DHCR7	100	100	100	100	Smith-Lemli-Opitz syndrome, 270400
DKC1	99,7	97,2	100	99,6	Dyskeratosis congenita, X-linked, 305000
DNAJB11	99,9	99,6	100	100	Polycystic kidney disease 6 with or without polycystic liver disease, 618061
EDNRB	96,3	92,5	100	100	ABCD syndrome, 600501 Waardenburg syndrome, type 4A, 277580

EPHX1	99,8	97,8	100	100	No OMIM disease ID
ETFDH	99,8	99,4	100	100	Glutaric acidemia IIC, 231680
FAH	100	99,5	100	99,9	Tyrosinemia, type I, 276700
FECH	99,9	99,8	100	100	Protoporphyria, erythropoietic, 1, 177000
FH	93,2	87,2	100	100	Leiomyomatosis and renal cell cancer, 150800 Fumarase deficiency, 606812
FLNA	100	99,9	100	100	Otopalatodigital syndrome, type II, 304120 Intestinal pseudoobstruction, neuronal, 300048 Cardiac valvular dysplasia, X-linked, 314400 ?FG syndrome 2, 300321 Melnick-Needles syndrome, 309350 Terminal osseous dysplasia, 300244 Congenital short bowel syndrome, 300048 Otopalatodigital syndrome, type I, 311300 Heterotopia, periventricular, 1, 300049 Frontometaphyseal dysplasia 1, 305620
GALT	100	99,6	100	100	Galactosemia, 230400
GANAB	99,8	97,8	100	100	Polycystic kidney disease 3, 600666
GBA	100	100	100	100	Gaucher disease, type II, 230900 Gaucher disease, type IIIC, 231005 Gaucher disease, type III, 231000 Gaucher disease, type I, 230800 Gaucher disease, perinatal lethal, 608013
GBE1	99,9	99,7	100	100	Glycogen storage disease IV, 232500 Polyglucosan body disease, adult form, 263570
GDNF	100	100	100	100	No OMIM disease ID
GFM1	99,7	98,7	100	100	Combined oxidative phosphorylation deficiency 1, 609060
GLI3	98,5	97,7	100	100	Greig cephalopolysyndactyly syndrome, 175700 Polydactyly, postaxial, types A1 and B, 174200 Pallister-Hall syndrome, 146510 Polydactyly, preaxial, type IV, 174700
GLIS3	98,5	97,4	100	100	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199
HADHA	95,5	88,3	100	100	HELLP syndrome, maternal, of pregnancy, 609016 Mitochondrial trifunctional protein deficiency, 609015

					LCHAD deficiency, 609016 Fatty liver, acute, of pregnancy, 609016
HAMP	100	100	100	100	Hemochromatosis, type 2B, 613313
HFE	99,9	97,8	100	100	Hemochromatosis, 235200
HNF1B	99	95,7	100	100	Type 2 diabetes mellitus, 125853 Renal cysts and diabetes syndrome, 137920
HSD17B4	95,3	92,8	96,6	96,6	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
HSD3B7	98,9	95	100	100	Bile acid synthesis defect, congenital, 1, 607765
IARS1	99,9	99,4	100	100	Growth retardation, impaired intellectual development, hypotonia, and hepatopathy, 617093
IFT140	99,9	99,2	100	100	Short-rib thoracic dysplasia 9 with or without polydactyly, 266920 Retinitis pigmentosa 80, 617781
IFT172	99,6	98,6	100	100	Retinitis pigmentosa 71, 616394 Bardet-Biedl syndrome 20, 619471 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT43	100	100	100	100	?Cranioectodermal dysplasia 3, 614099 ?Retinitis pigmentosa 81, 617871 Short-rib thoracic dysplasia 18 with polydactyly, 617866
INSR	97,3	93	100	99,6	Rabson-Mendenhall syndrome, 262190 Leprechaunism, 246200 Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549 Hyperinsulinemic hypoglycemia, familial, 5, 609968
INVS	100	99,8	100	100	Nephronophthisis 2, infantile, 602088
JAG1	97,8	96,7	100	100	?Deafness, congenital heart defects, and posterior embryotoxon, 617992 Alagille syndrome 1, 118450 Tetralogy of Fallot, 187500
LARS1	99,4	97,2	100	99,9	?Infantile liver failure syndrome 1, 615438
LRP5	99,2	98,2	99,8	99,2	Osteopetrosis, autosomal dominant 1, 607634 Hyperostosis, endosteal, 144750 Osteosclerosis, 144750 Polycystic liver disease 4 with or without kidney cysts, 617875 Osteoporosis-pseudoglioma syndrome, 259770 Exudative vitreoretinopathy 4, 601813 van Buchem disease, type 2, 607636
MARS1	99	96,1	100	100	Interstitial lung and liver disease, 615486 Charcot-Marie-Tooth disease, axonal, type 2U, 616280

MPV17	100	98,7	100	100	Charcot-Marie-Tooth disease, axonal, type 2EE, 618400 Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810
MTM1	98,7	92	100	99,7	Myotubular myopathy, X-linked, 310400
MYO5B	98,5	94,8	100	100	Diarrhea 2, with microvillus atrophy, 251850
NBAS	99,9	99,3	100	100	Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800 Infantile liver failure syndrome 2, 616483
NHP2	100	100	100	100	Dyskeratosis congenita, autosomal recessive 2, 613987
NOP10	100	99,2	100	100	Dyskeratosis congenita, autosomal recessive 1, 224230
NOTCH2	100	99,2	100	100	Alagille syndrome 2, 610205 Hajdu-Cheney syndrome, 102500
NPC1	99,9	99	100	100	Niemann-Pick disease, type C1, 257220 Niemann-Pick disease, type D, 257220
NPC2	100	99,2	100	100	Niemann-pick disease, type C2, 607625
NPHP3	99,6	98,5	100	99,9	Nephronophthisis 3, 604387 Renal-hepatic-pancreatic dysplasia 1, 208540 Meckel syndrome 7, 267010
NR1H4	99,6	98,6	100	100	Cholestasis, progressive familial intrahepatic, 5, 617049
PEX1	99,8	99,4	100	100	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 Peroxisome biogenesis disorder 1A (Zellweger), 214100
PEX10	98,8	90,6	100	100	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871
PEX12	100	100	100	100	Peroxisome biogenesis disorder 3B, 266510 Peroxisome biogenesis disorder 3A (Zellweger), 614859
PEX13	100	100	100	100	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885
PEX14	95,8	89,4	100	100	Peroxisome biogenesis disorder 13A (Zellweger), 614887
PEX16	97,1	93,9	100	100	Peroxisome biogenesis disorder 8B, 614877 Peroxisome biogenesis disorder 8A (Zellweger), 614876
PEX19	99	94,4	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	99,8	100	100	Peroxisome biogenesis disorder 7B, 614873 Peroxisome biogenesis disorder 7A (Zellweger), 614872

PEX3	99,4	99,2	100	100	Peroxisome biogenesis disorder 10A (Zellweger), 614882 ?Peroxisome biogenesis disorder 10B, 617370
PEX5	99,9	98,8	100	100	Peroxisome biogenesis disorder 2B, 202370 Peroxisome biogenesis disorder 2A (Zellweger), 214110 Rhizomelic chondrodysplasia punctata, type 5, 616716
PEX6	96,4	88	100	100	Peroxisome biogenesis disorder 4B, 614863 Peroxisome biogenesis disorder 4A (Zellweger), 614862 Heimler syndrome 2, 616617
PEX7	88	81	91,3	91,2	Rhizomelic chondrodysplasia punctata, type 1, 215100 Peroxisome biogenesis disorder 9B, 614879
PKD1	40,6	32,8	99,3	99	Polycystic kidney disease 1, 173900
PKD2	96	93,3	99,6	97,9	Polycystic kidney disease 2, 613095
PKHD1	100	99,6	100	100	Polycystic kidney disease 4, with or without hepatic disease, 263200
POLG	99,9	98,8	100	100	Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 Progressive external ophthalmoplegia, autosomal dominant 1, 157640 Progressive external ophthalmoplegia, autosomal recessive 1, 258450
POMC	100	100	100	100	Obesity, adrenal insufficiency, and red hair due to POMC deficiency, 609734
PRKCSH	99,5	94,1	100	100	Polycystic liver disease 1, 174050
RAD21	99,2	95,9	100	100	Cornelia de Lange syndrome 4, 614701 ?Mungan syndrome, 611376
RFX6	99,9	99,5	100	100	Mitchell-Riley syndrome, 615710
RINT1	99,6	97,6	100	99,9	Infantile liver failure syndrome 3, 618641
RPGRIP1L	96,5	95,3	100	99,4	Joubert syndrome 7, 611560 Meckel syndrome 5, 611561 ?COACH syndrome 3, 619113
SC5D	99,9	99,1	100	100	Lathosterolosis, 607330
SCO1	97,6	94,4	100	100	Mitochondrial complex IV deficiency, nuclear type 4, 619048
SCYL1	100	99,9	100	100	Spinocerebellar ataxia, autosomal recessive 21, 616719
SEC61B	97,4	89,1	100	100	No OMIM disease ID
SEC63	86,2	77,3	100	100	Polycystic liver disease 2, 617004

SERPINA1	100	100	100	100	Hemorrhagic diathesis due to antithrombin Pittsburgh, 613490 Emphysema due to AAT deficiency, 613490 Emphysema-cirrhosis, due to AAT deficiency, 613490
SGO1	99,5	99	100	100	Chronic atrial and intestinal dysrhythmia, 616201
SLC25A13	100	99,4	100	100	Citrullinemia, type II, neonatal-onset, 605814 Citrullinemia, adult-onset type II, 603471
SLC40A1	99,9	98,6	100	99,9	Hemochromatosis, type 4, 606069
SMPD1	100	99,9	100	100	Niemann-Pick disease, type B, 607616 Niemann-Pick disease, type A, 257200
STN1	99,9	99,8	100	100	Cerebroretinal microangiopathy with calcifications and cysts 2, 617341
TALDO1	100	98	100	100	Transaldolase deficiency, 606003
TERC	0	0	0	0	Dyskeratosis congenita, autosomal dominant 1, 127550
TERT	97	94,8	100	100	No OMIM disease ID
TFR2	99,3	96,9	100	100	Hemochromatosis, type 3, 604250
TJP2	92,8	92,3	98,8	98,8	Hypercholanemia, familial 1, 607748 Cholestasis, progressive familial intrahepatic 4, 615878
TMEM67	98,6	93,5	100	99,6	Nephronophthisis 11, 613550 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361 ?RHYNS syndrome, 602152 COACH syndrome 1, 216360
TRAF3IP1	98,7	95,4	100	100	Senior-Loken syndrome 9, 616629
TRMU	99,9	99,6	100	99,9	Liver failure, transient infantile, 613070
TTC37	99,7	98,8	100	100	Trichohepatoenteric syndrome 1, 222470
TWNK	100	99,9	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	99,4	100	100	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041
UBR1	99,6	99,1	98	97,9	Johanson-Blizzard syndrome, 243800
UGT1A1	100	100	100	100	Crigler-Najjar syndrome, type I, 218800 Hyperbilirubinemia, familial transient neonatal, 237900 Crigler-Najjar syndrome, type II, 606785
VIPAS39	100	100	100	100	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VPS33B	100	99,9	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.
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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 : September 16th , 2021.
This list is accurate for panel version DG 3.2.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions betw een {} signify risk factors