LIVER DISORDERS GENE PANEL DG 2.18 (126 genes)

Releasedate: 20-04-2020

Gene	Agilent V5 covered >10x	Agilent V5 covered > 20x	TWIST covered >10x	TWIST covered >20x	Associated Phenotype description and OMIM disease ID
ABCB11	100%	99,70%	100%	100%	Cholestasis, progressive familial intrahepatic 2, 601847 Cholestasis, benign recurrent intrahepatic, 2, 605479
ABCB4	99,90%	99,60%	100%	100%	Gallbladder disease 1, 600803 Cholestasis, intrahepatic, of pregnancy, 3, 614972 Cholestasis, progressive familial intrahepatic 3, 602347
ABCC2	100%	99,90%	100%	100%	Dubin-Johnson syndrome, 237500
ABCD3	99,80%	97,70%	100%	100%	?Bile acid synthesis defect, congenital, 5, 616278
ACOX2	100%	99,20%	100%	100%	Bile acid synthesis defect, congenital, 6, 617308
ACTA2	100%	99,00%	100%	100%	Aortic aneurysm, familial thoracic 6, 611788 Multisystemic smooth muscle dysfunction syndrome, 613834 Moyamoya disease 5, 614042
ACTG2	99,90%	98,20%	100%	100%	Visceral myopathy, 155310
ADK	99,50%	95,80%	100%	100%	Hypermethioninemia due to adenosine kinase deficiency, 614300
AHCY	100%	99,20%	100%	100%	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752
AKR1D1	100%	99,40%	100%	100%	Bile acid synthesis defect, congenital, 2, 235555
ALDOB	100%	99,10%	100%	100%	Fructose intolerance, hereditary, 229600
ALG8	97,20%	95,60%	96,60%	96,60%	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	93,80%	89,50%	97,90%	95,80%	Nephronophthisis 16, 615382
AP1S1	99,90%	99,50%	100%	100%	MEDNIK syndrome, 609313
ATP7B	99,90%	99,20%	100%	100%	Wilson disease, 277900
ATP8B1	96,50%	94,00%	100%	100%	Cholestasis, intrahepatic, of pregnancy, 1, 147480 Cholestasis, progressive familial intrahepatic 1, 211600 Cholestasis, benign recurrent intrahepatic, 243300
BAAT	99,80%	98,40%	100%	100%	Hypercholanemia, familial, 607748
BCS1L	100%	100%	100%	100%	Leigh syndrome, 256000 GRACILE syndrome, 603358

					Bjornstad syndrome, 262000 Mitochondrial complex III deficiency, nuclear type 1, 124000
BLVRA	100%	99,40%	100%	100%	Hyperbiliverdinemia, 614156
CC2D2A	99,70%	97,70%	98,20%	98,20%	Meckel syndrome 6, 612284 Joubert syndrome 9, 612285 COACH syndrome, 216360
CEP83	99,80%	97,40%	100%	100%	Nephronophthisis 18, 615862
CFC1	84,20%	74,10%	100%	100%	Heterotaxy, visceral, 2, autosomal, 605376
CFTR	99,60%	97,90%	100%	100%	Cystic fibrosis, 219700 Congenital bilateral absence of vas deferens, 277180 Sweat chloride elevation without CF, 0
CHD8	100%	99,90%	100%	100%	No OMIM disease ID
CHRM3	100%	100%	100%	100%	?Prune belly syndrome, 100100
CHRNA3	100%	99,40%	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%	99,60%	100%	100%	Congenital short bowel syndrome, 615237
COG7	100%	100%	100%	100%	Congenital disorder of glycosylation, type IIe, 608779
CYP27A1	98,90%	96,70%	100%	100%	Cerebrotendinous xanthomatosis, 213700
CYP7B1	98,00%	92,80%	100%	100%	Spastic paraplegia 5A, autosomal recessive, 270800 Bile acid synthesis defect, congenital, 3, 613812
DCDC2	100%	99,90%	100%	100%	Sclerosing cholangitis, neonatal, 617394 Nephronophthisis 19, 616217 ?Deafness, autosomal recessive 66, 610212
DGUOK	100%	99,40%	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	99,80%	98,70%	100%	99,70%	Dyskeratosis congenita, X-linked, 305000
DNAJB11	100%	99,50%	100%	100%	Polycystic kidney disease 6 with or without polycystic liver disease, 618061
EDNRB	98,00%	93,80%	100%	100%	Waardenburg syndrome, type 4A, 277580 ABCD syndrome, 600501
EPHX1	99,90%	98,80%	100%	100%	?Hypercholanemia, familial, 607748
ETFDH	100%	99,80%	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	92,10%	88,30%	100%	100%	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800

FLNA	100%	99,90%	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%	99,70%	100%	100%	Galactosemia, 230400
GANAB	99,90%	99,00%	100%	100%	Polycystic kidney disease 3, 600666
GBA	100%	100%	100%	100%	Gaucher disease, type III, 231000 Gaucher disease, type IIIC, 231005 Gaucher disease, type I, 230800 Gaucher disease, perinatal lethal, 608013 Gaucher disease, type II, 230900
GBE1	100%	99,60%	100%	100%	Polyglucosan body disease, adult form, 263570 Glycogen storage disease IV, 232500
GDNF	100%	100%	100%	100%	Central hypoventilation syndrome, 209880
GFM1	99,90%	99,40%	100%	100%	Combined oxidative phosphorylation deficiency 1, 609060
GLI3	100%	99,50%	100%	100%	Polydactyly, postaxial, types A1 and B, 174200 Greig cephalopolysyndactyly syndrome, 175700 Polydactyly, preaxial, type IV, 174700 Pallister-Hall syndrome, 146510
GLIS3	100%	99,60%	100%	100%	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199
HADHA	97,10%	91,30%	100%	100%	LCHAD deficiency, 609016 HELLP syndrome, maternal, of pregnancy, 609016 Fatty liver, acute, of pregnancy, 609016 Trifunctional protein deficiency, 609015
HAMP	100%	100%	100%	100%	Hemochromatosis, type 2B, 613313
HFE	100%	99,70%	100%	100%	Hemochromatosis, 235200
HNF1B	99,30%	96,10%	100%	100%	Renal cysts and diabetes syndrome, 137920 Diabetes mellitus, noninsulin-dependent, 125853
HSD17B4	96,00%	93,70%	96,60%	96,60%	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
HSD3B7	99,10%	95,50%	100%	100%	Bile acid synthesis defect, congenital, 1, 607765
IARS	100%	99,60%	100%	100%	Growth retardation, impaired intellectual development, hypotonia, and hepatopathy, 617093

IFT140	99,80%	98,80%	100%	100%	Retinitis pigmentosa 80, 617781 Short-rib thoracic dysplasia 9 with or without polydactyly, 266920
IFT172	99,90%	99,10%	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	97,80%	94,70%	99,90%	99,20%	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	97,70%	96,80%	100%	100%	Alagille syndrome 1, 118450 Tetralogy of Fallot, 187500 ?Deafness, congenital heart defects, and posterior embryotoxon, 617992
LARS	99,80%	98,40%	100%	100%	?Infantile liver failure syndrome 1, 615438
LRP5	98,50%	98,10%	100%	99,70%	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
MARS	99,70%	97,40%	100%	100%	Interstitial lung and liver disease, 615486 Charcot-Marie-Tooth disease, axonal, type 2U, 616280
MPV17	100%	97,20%	100%	100%	Charcot-Marie-Tooth disease, axonal, type 2EE, 618400 Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810
MTM1	99,00%	93,30%	100%	100%	Myotubular myopathy, X-linked, 310400
MYO5B	99,10%	96,20%	100%	100%	Microvillus inclusion disease, 251850
NBAS	100%	99,60%	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%	99,80%	100%	100%	Dyskeratosis congenita, autosomal recessive 1, 224230
NOTCH2	100%	99,50%	100%	100%	Hajdu-Cheney syndrome, 102500 Alagille syndrome 2, 610205
NPC1	99,60%	98,70%	100%	100%	Niemann-Pick disease, type D, 257220 Niemann-Pick disease, type C1, 257220
NPC2	100%	99,60%	100%	100%	Niemann-pick disease, type C2, 607625

NPHP3	99,70%	98,40%	100%	100%	Meckel syndrome 7, 267010 Renal-hepatic-pancreatic dysplasia 1, 208540 Nephronophthisis 3, 604387
NR1H4	99,80%	98,50%	100%	100%	Cholestasis, progressive familial intrahepatic, 5, 617049
PEX1	99,90%	99,40%	100%	100%	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 Peroxisome biogenesis disorder 1A (Zellweger), 214100
PEX10	96,80%	89,70%	100%	99,90%	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	96,70%	90,80%	100%	100%	Peroxisome biogenesis disorder 13A (Zellweger), 614887
PEX16	97,90%	94,20%	100%	100%	Peroxisome biogenesis disorder 8A (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877
PEX19	99,90%	98,50%	100%	100%	Peroxisome biogenesis disorder 12A (Zellweger), 614886
PEX2	100%	100%	100%	100%	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867
PEX26	100%	100%	100%	100%	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873
PEX3	100%	99,30%	100%	100%	Peroxisome biogenesis disorder 10A (Zellweger), 614882 ?Peroxisome biogenesis disorder 10B, 617370
PEX5	99,90%	99,00%	100%	100%	Peroxisome biogenesis disorder 2B, 202370 Rhizomelic chondrodysplasia punctata, type 5, 616716 Peroxisome biogenesis disorder 2A (Zellweger), 214110
PEX6	94,50%	86,70%	100%	100%	Peroxisome biogenesis disorder 4B, 614863 Heimler syndrome 2, 616617 Peroxisome biogenesis disorder 4A (Zellweger), 614862
PEX7	87,80%	80,70%	91,30%	91,30%	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100
TALDO1	100%	97,90%	100%	100%	Transaldolase deficiency, 606003
PKD1	39,20%	30,00%	99,20%	98,90%	Polycystic kidney disease 1, 173900
PKD2	95,50%	91,10%	99,30%	97,70%	Polycystic kidney disease 2, 613095
PKHD1	100%	99,60%	100%	100%	Polycystic kidney disease 4, with or without hepatic disease, 263200
POLG	100%	99,30%	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, adrenal insufficiency, and red hair due to POMC deficiency, 609734
PRKCSH	99,80%	95,40%	100%	100%	Polycystic liver disease 1, 174050
RAD21	99,20%	96,60%	100%	100%	?Mungan syndrome, 611376 Cornelia de Lange syndrome 4, 614701
RFX6	100%	99,60%	100%	100%	Mitchell-Riley syndrome, 615710
RPGRIP1L	96,70%	95,70%	100%	99,50%	COACH syndrome, 216360 Meckel syndrome 5, 611561 Joubert syndrome 7, 611560
SC5D	100%	99,50%	100%	100%	Lathosterolosis, 607330
SCO1	97,10%	93,80%	100%	100%	Mitochondrial complex IV deficiency, 220110
SEC61B	99,10%	92,40%	100%	100%	No OMIM disease ID
SEC63	91,20%	83,30%	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	99,90%	98,90%	100%	100%	Chronic atrial and intestinal dysrhythmia, 616201
SLC25A13	100%	99,70%	100%	100%	Citrullinemia, adult-onset type II, 603471 Citrullinemia, type II, neonatal-onset, 605814
SLC40A1	100%	99,50%	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
TERC	NC	NC	NC	NC	Dyskeratosis congenita, autosomal dominant 1, 127550
TERT	96,20%	94,50%	100%	100%	No OMIM disease ID
TFR2	99,10%	97,80%	100%	100%	Hemochromatosis, type 3, 604250
TJP2	94,00%	93,60%	100%	100%	Hypercholanemia, familial, 607748 Cholestasis, progressive familial intrahepatic 4, 615878
TMEM67	99,50%	95,00%	100%	99,90%	Meckel syndrome 3, 607361 ?RHYNS syndrome, 602152 Nephronophthisis 11, 613550 COACH syndrome, 216360 Joubert syndrome 6, 610688
TRAF3IP1	99,60%	97,60%	100%	100%	Senior-Loken syndrome 9, 616629
TRMU	100%	100%	100%	99,90%	Liver failure, transient infantile, 613070
TTC37	100%	99,30%	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%	97,00%	100%	100%	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041
UBR1	99,90%	99,10%	98,00%	98,00%	Johanson-Blizzard syndrome, 243800
UGT1A1	100%	100%	100%	100%	Hyperbilirubinemia, familial transient neonatal, 237900 Crigler-Najjar syndrome, type I, 218800 Crigler-Najjar syndrome, type II, 606785
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-DNA coding genes.
non-DNA coding genes are covered, but as coverage statistics are based on DNA coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : April 20th , 2020.
This list is accurate for panel version DG 2.18
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors