LIVER DISORDERS GENE PANEL DG 3.4.0 (128 genes)

Releasedate: 19-04-2022

Gene	*TWIST covered >10x*	*TWIST covered >20x*	*Associated Phenotype description and OMIM disease ID*
ABCB11	100,0%	100,0%	Cholestasis, benign recurrent intrahepatic, 2, 605479 Cholestasis, progressive familial intrahepatic 2, 601847
ABCB4	100,0%	100,0%	Gallbladder disease 1, 600803 Cholestasis, intrahepatic, of pregnancy, 3, 614972 Cholestasis, progressive familial intrahepatic 3, 602347
ABCC2	100,0%	100,0%	Dubin-Johnson syndrome, 237500
ABCD3	100,0%	100,0%	?Bile acid synthesis defect, congenital, 5, 616278
ACOX2	100,0%	100,0%	Bile acid synthesis defect, congenital, 6, 617308
ACTA2	100,0%	100,0%	Multisystemic smooth muscle dysfunction syndrome, 613834 Aortic aneurysm, familial thoracic 6, 611788 Moyamoya disease 5, 614042
ACTG2	100,0%	100,0%	Megacystis-microcolon-intestinal hypoperistalsis syndrome 5, 619431 Visceral myopathy 1, 155310
ADK	84,5%	84,5%	Hypermethioninemia due to adenosine kinase deficiency, 614300
AHCY	100,0%	100,0%	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752
AKR1D1	100,0%	100,0%	Bile acid synthesis defect, congenital, 2, 235555
ALDOB	100,0%	100,0%	Fructose intolerance, hereditary, 229600
ALG8	96,6%	96,6%	Congenital disorder of glycosylation, type Ih, 608104 Polycystic liver disease 3 with or without kidney cysts, 617874
AMACR	100,0%	100,0%	Alpha-methylacyl-CoA racemase deficiency, 614307 Bile acid synthesis defect, congenital, 4, 214950
ANKS6	100,0%	99,9%	Nephronophthisis 16, 615382
AP1S1	100,0%	100,0%	MEDNIK syndrome, 609313
ATP7B	100,0%	100,0%	Wilson disease, 277900
ATP8B1	100,0%	100,0%	Cholestasis, progressive familial intrahepatic 1, 211600 Cholestasis, intrahepatic, of pregnancy, 1, 147480 Cholestasis, benign recurrent intrahepatic, 243300
BAAT	100,0%	100,0%	Bile acid conjugation defect 1, 619232

BCS1L	100,0%	100,0%	GRACILE syndrome, 603358 Mitochondrial complex III deficiency, nuclear type 1, 124000 Bjornstad syndrome, 262000
BLVRA	100,0%	100,0%	Hyperbiliverdinemia, 614156
CC2D2A	97,1%	97,1%	COACH syndrome 2, 619111 Meckel syndrome 6, 612284 Joubert syndrome 9, 612285
CEP83	100,0%	100,0%	Nephronophthisis 18, 615862
CFC1	100,0%	100,0%	Heterotaxy, visceral, 2, autosomal, 605376
CFTR	100,0%	100,0%	Cystic fibrosis, 219700 Congenital bilateral absence of vas deferens, 277180 Sweat chloride elevation without CF,
CHD8	100,0%	100,0%	No OMIM Disease ID
CHRM3	100,0%	100,0%	Prune belly syndrome, 100100
CHRNA3	100,0%	100,0%	Bladder dysfunction, autonomic, with impaired pupillary reflex and secondary CAKUT, 191800
CLDN1	100,0%	100,0%	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis, 607626
CLMP	100,0%	100,0%	Congenital short bowel syndrome, 615237
COG7	100,0%	100,0%	Congenital disorder of glycosylation, type IIe, 608779
CYP27A1	100,0%	100,0%	Cerebrotendinous xanthomatosis, 213700
CYP7B1	100,0%	100,0%	Spastic paraplegia 5A, autosomal recessive, 270800 Bile acid synthesis defect, congenital, 3, 613812
DCDC2	100,0%	100,0%	Nephronophthisis 19, 616217 ?Deafness, autosomal recessive 66, 610212 Sclerosing cholangitis, neonatal, 617394
DGUOK	100,0%	100,0%	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	100,0%	100,0%	Smith-Lemli-Opitz syndrome, 270400
DKC1	100,0%	100,0%	Dyskeratosis congenita, X-linked, 305000
DNAJB11	100,0%	100,0%	Polycystic kidney disease 6 with or without polycystic liver disease, 618061
EDNRB	100,0%	100,0%	ABCD syndrome, 600501 Waardenburg syndrome, type 4A, 277580
EPHX1	100,0%	100,0%	No OMIM Disease ID
ETFDH	100,0%	100,0%	Glutaric acidemia IIC, 231680
FAH	100,0%	100,0%	Tyrosinemia, type I, 276700
FECH	100,0%	100,0%	Protoporphyria, erythropoietic, 1, 177000

FH	100,0%	100,0%	Leiomyomatosis and renal cell cancer, 150800 Fumarase deficiency, 606812
FLNA	100,0%	100,0%	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	100,0%	100,0%	Galactosemia, 230400
GANAB	100,0%	100,0%	Polycystic kidney disease 3, 600666
GBA	100,0%	100,0%	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	100,0%	100,0%	Glycogen storage disease IV, 232500 Polyglucosan body disease, adult form, 263570
GDNF	100,0%	100,0%	No OMIM Disease ID
GFM1	100,0%	100,0%	Combined oxidative phosphorylation deficiency 1, 609060
GLI3	100,0%	100,0%	Greig cephalopolysyndactyly syndrome, 175700 Polydactyly, postaxial, types A1 and B, 174200 Pallister-Hall syndrome, 146510 Polydactyly, preaxial, type IV, 174700
GLIS3	100,0%	100,0%	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199
HADHA	100,0%	100,0%	HELLP syndrome, maternal, of pregnancy, 609016 Mitochondrial trifunctional protein deficiency, 609015 LCHAD deficiency, 609016 Fatty liver, acute, of pregnancy, 609016
HAMP	100,0%	100,0%	Hemochromatosis, type 2B, 613313
HFE	100,0%	100,0%	Hemochromatosis, 235200
HNF1B	100,0%	100,0%	Type 2 diabetes mellitus, 125853 Renal cysts and diabetes syndrome, 137920
HSD17B4	96,6%	96,6%	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400

HSD3B7	100,0%	100,0%	Bile acid synthesis defect, congenital, 1, 607765
IARS1	100,0%	100,0%	Growth retardation, impaired intellectual development, hypotonia, and hepatopathy, 617093
IFT140	100,0%	100,0%	Short-rib thoracic dysplasia 9 with or without polydactyly, 266920 Retinitis pigmentosa 80, 617781
IFT172	100,0%	100,0%	Retinitis pigmentosa 71, 616394 Bardet-Biedl syndrome 20, 619471 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT43	100,0%	100,0%	?Cranioectodermal dysplasia 3, 614099 ?Retinitis pigmentosa 81, 617871 Short-rib thoracic dysplasia 18 with polydactyly, 617866
INSR	100,0%	100,0%	Rabson-Mendenhall syndrome, 262190 Leprechaunism, 246200 Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549 Hyperinsulinemic hypoglycemia, familial, 5, 609968
INVS	100,0%	100,0%	Nephronophthisis 2, infantile, 602088
JAG1	100,0%	100,0%	?Deafness, congenital heart defects, and posterior embryotoxon, 617992 Charcot-Marie-Tooth disease, axonal, type 2HH, 619574 Alagille syndrome 1, 118450 Tetralogy of Fallot, 187500
LARS1	100,0%	100,0%	?Infantile liver failure syndrome 1, 615438
LRP5	100,0%	100,0%	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	100,0%	100,0%	Interstitial lung and liver disease, 615486 ?Trichothiodystrophy 9, nonphotosensitive, 619692 Charcot-Marie-Tooth disease, axonal, type 2U, 616280
MPV17	100,0%	100,0%	Charcot-Marie-Tooth disease, axonal, type 2EE, 618400 Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810
MTM1	100,0%	100,0%	Myopathy, centronuclear, X-linked, 310400
MYO5B	100,0%	100,0%	Diarrhea 2, with microvillus atrophy, 251850
NBAS	100,0%	100,0%	Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800 Infantile liver failure syndrome 2, 616483
NHP2	100,0%	100,0%	Dyskeratosis congenita, autosomal recessive 2, 613987
NOP10	100,0%	100,0%	Dyskeratosis congenita, autosomal recessive 1, 224230

NOTCH2	100,0%	100,0%	Alagille syndrome 2, 610205 Hajdu-Cheney syndrome, 102500
NPC1	100,0%	100,0%	Niemann-Pick disease, type C1, 257220 Niemann-Pick disease, type D, 257220
NPC2	100,0%	100,0%	Niemann-pick disease, type C2, 607625
NPHP3	100,0%	100,0%	Nephronophthisis 3, 604387 Renal-hepatic-pancreatic dysplasia 1, 208540 Meckel syndrome 7, 267010
NR1H4	100,0%	100,0%	Cholestasis, progressive familial intrahepatic, 5, 617049
PEX1	100,0%	100,0%	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 Peroxisome biogenesis disorder 1A (Zellweger), 214100
PEX10	100,0%	100,0%	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871
PEX12	100,0%	100,0%	Peroxisome biogenesis disorder 3B, 266510 Peroxisome biogenesis disorder 3A (Zellweger), 614859
PEX13	100,0%	100,0%	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885
PEX14	100,0%	100,0%	Peroxisome biogenesis disorder 13A (Zellweger), 614887
PEX16	100,0%	100,0%	Peroxisome biogenesis disorder 8B, 614877 Peroxisome biogenesis disorder 8A (Zellweger), 614876
PEX19	100,0%	100,0%	Peroxisome biogenesis disorder 12A (Zellweger), 614886
PEX2	100,0%	100,0%	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867
PEX26	100,0%	100,0%	Peroxisome biogenesis disorder 7B, 614873 Peroxisome biogenesis disorder 7A (Zellweger), 614872
PEX3	100,0%	100,0%	Peroxisome biogenesis disorder 10A (Zellweger), 614882 ?Peroxisome biogenesis disorder 10B, 617370
PEX5	100,0%	100,0%	Peroxisome biogenesis disorder 2B, 202370 Peroxisome biogenesis disorder 2A (Zellweger), 214110 Rhizomelic chondrodysplasia punctata, type 5, 616716
PEX6	100,0%	100,0%	Peroxisome biogenesis disorder 4B, 614863 Peroxisome biogenesis disorder 4A (Zellweger), 614862 Heimler syndrome 2, 616617
PEX7	91,3%	91,3%	Rhizomelic chondrodysplasia punctata, type 1, 215100 Peroxisome biogenesis disorder 9B, 614879
PKD1	99,9%	99,7%	Polycystic kidney disease 1, 173900
PKD2	100,0%	100,0%	Polycystic kidney disease 2, 613095

PKHD1	100,0%	100,0%	Polycystic kidney disease 4, with or without hepatic disease, 263200
POLG	100,0%	100,0%	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	100,0%	100,0%	Obesity, adrenal insufficiency, and red hair due to POMC deficiency, 609734
PRKCSH	100,0%	100,0%	Polycystic liver disease 1, 174050
RAD21	100,0%	100,0%	Cornelia de Lange syndrome 4, 614701 ?Mungan syndrome, 611376
RFX6	100,0%	100,0%	Mitchell-Riley syndrome, 615710
RINT1	100,0%	100,0%	Infantile liver failure syndrome 3, 618641
RPGRIP1L	100,0%	99,8%	Joubert syndrome 7, 611560 Meckel syndrome 5, 611561 ?COACH syndrome 3, 619113
SC5D	100,0%	100,0%	Lathosterolosis, 607330
SCO1	100,0%	100,0%	Mitochondrial complex IV deficiency, nuclear type 4, 619048
SCYL1	100,0%	100,0%	Spinocerebellar ataxia, autosomal recessive 21, 616719
SEC61B	100,0%	100,0%	No OMIM Disease ID
SEC63	100,0%	100,0%	Polycystic liver disease 2, 617004
SERPINA1	100,0%	100,0%	Hemorrhagic diathesis due to antithrombin Pittsburgh, 613490 Emphysema due to AAT deficiency, 613490 Emphysema-cirrhosis, due to AAT deficiency, 613490
SGO1	100,0%	100,0%	Chronic atrial and intestinal dysrhythmia, 616201
SLC25A13	100,0%	100,0%	Citrullinemia, type II, neonatal-onset, 605814 Citrullinemia, adult-onset type II, 603471
SLC40A1	100,0%	100,0%	Hemochromatosis, type 4, 606069
SMPD1	100,0%	100,0%	Niemann-Pick disease, type B, 607616 Niemann-Pick disease, type A, 257200
STN1	100,0%	100,0%	Cerebroretinal microangiopathy with calcifications and cysts 2, 617341
TALDO1	100,0%	100,0%	Transaldolase deficiency, 606003
TERC	NC	NC	Dyskeratosis congenita, autosomal dominant 1, 127550
TERT	100,0%	100,0%	Dyskeratosis congenita, autosomal dominant 2, 613989 Dyskeratosis congenita, autosomal recessive 4, 613989 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1, 614742
TFR2	100,0%	100,0%	Hemochromatosis, type 3, 604250

TJP2	98,8%	98,8%	Hypercholanemia, familial 1, 607748 Cholestasis, progressive familial intrahepatic 4, 615878
TMEM67	100,0%	100,0%	Nephronophthisis 11, 613550 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361 ?RHYNS syndrome, 602152 COACH syndrome 1, 216360
TRAF3IP1	100,0%	100,0%	Senior-Loken syndrome 9, 616629
TRMU	100,0%	100,0%	Liver failure, transient infantile, 613070
TTC37	100,0%	100,0%	Trichohepatoenteric syndrome 1, 222470
TWNK	100,0%	100,0%	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286 Perrault syndrome 5, 616138
TYMP	100,0%	100,0%	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041
UBR1	98,0%	98,0%	Johanson-Blizzard syndrome, 243800
UGT1A1	100,0%	100,0%	Crigler-Najjar syndrome, type I, 218800 Hyperbilirubinemia, familial transient neonatal, 237900 Crigler-Najjar syndrome, type II, 606785
VIPAS39	100,0%	100,0%	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VPS33B	100,0%	100,0%	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.
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.
TWIST is the chemistry used for 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 : April 19th , 2022.
This list is accurate for panel version DG 3.4.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors