LIVER DISORDERS GENE PANEL DG 2.17 (113 genes)

Releasedate: 06-12-2019

Gene	Median Coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM disease ID
ABCB11	135.9	100.0%	99.4%	Cholestasis, progressive familial intrahepatic 2, 601847 Cholestasis, benign recurrent intrahepatic, 2, 605479
ABCB4	125.0	100.0%	99.1%	Gallbladder disease 1, 600803 Cholestasis, intrahepatic, of pregnancy, 3, 614972 Cholestasis, progressive familial intrahepatic 3, 602347
ABCC2	113.1	100.0%	99.8%	Dubin-Johnson syndrome, 237500
ABCD3	106.9	99.6%	96.7%	?Bile acid synthesis defect, congenital, 5, 616278
ACOX2	117.5	100.0%	99.6%	Bile acid synthesis defect, congenital, 6, 617308
ADK	101.3	99.9%	97.3%	Hypermethioninemia due to adenosine kinase deficiency, 614300
AHCY	120.8	100.0%	98.5%	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752
AKR1D1	92.8	99.2%	96.1%	Bile acid synthesis defect, congenital, 2, 235555
ALDOB	140.0	100.0%	99.1%	Fructose intolerance, hereditary, 229600
ALG8	118.5	96.8%	95.7%	Congenital disorder of glycosylation, type Ih, 608104 Polycystic liver disease 3 with or without kidney cysts, 617874
AMACR	168.4	100.0%	100.0%	Bile acid synthesis defect, congenital, 4, 214950 Alpha-methylacyl-CoA racemase deficiency, 614307
ANKS6	101.2	99.3%	96.5%	Nephronophthisis 16, 615382
AP1S1	105.8	100.0%	99.9%	MEDNIK syndrome, 609313
ATP7B	137.1	99.9%	99.3%	Wilson disease, 277900
ATP8B1	115.1	98.0%	95.0%	Cholestasis, intrahepatic, of pregnancy, 1, 147480 Cholestasis, progressive familial intrahepatic 1, 211600 Cholestasis, benign recurrent intrahepatic, 243300
BAAT	110.3	99.7%	97.9%	Hypercholanemia, familial, 607748
BCS1L	160.0	100.0%	100.0%	Leigh syndrome, 256000 GRACILE syndrome, 603358 Bjornstad syndrome, 262000 Mitochondrial complex III deficiency, nuclear type 1, 124000
BLVRA	117.6	100.0%	99.9%	Hyperbiliverdinemia, 614156

CC2D2A	112.6	99.0%	97.0%	Meckel syndrome 6, 612284 Joubert syndrome 9, 612285 COACH syndrome, 216360
CEP83	103.3	99.8%	96.2%	Nephronophthisis 18, 615862
CFC1	136.1	91.1%	82.3%	Heterotaxy, visceral, 2, autosomal, 605376
CFTR	112.9	99.4%	97.3%	Cystic fibrosis, 219700 Congenital bilateral absence of vas deferens, 277180 Sweat chloride elevation without CF, 0
CLDN1	129.7	100.0%	100.0%	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis, 607626
COG7	111.8	100.0%	99.9%	Congenital disorder of glycosylation, type IIe, 608779
CYP27A1	184.4	100.0%	99.8%	Cerebrotendinous xanthomatosis, 213700
CYP7B1	103.2	99.7%	97.2%	Spastic paraplegia 5A, autosomal recessive, 270800 Bile acid synthesis defect, congenital, 3, 613812
DCDC2	158.0	100.0%	99.9%	Sclerosing cholangitis, neonatal, 617394 Nephronophthisis 19, 616217 ?Deafness, autosomal recessive 66, 610212
DGUOK	127.0	100.0%	98.8%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, 617070 Portal hypertension, noncirrhotic, 617068 Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880
DHCR7	158.7	100.0%	100.0%	Smith-Lemli-Opitz syndrome, 270400
DKC1	93.9	99.7%	98.0%	Dyskeratosis congenita, X-linked, 305000
DNAJB11	107.4	99.9%	99.4%	Polycystic kidney disease 6 with or without polycystic liver disease, 618061
EPHX1	124.0	99.2%	96.3%	?Hypercholanemia, familial, 607748
ETFDH	112.7	100.0%	99.7%	Glutaric acidemia IIC, 231680
FAH	136.7	100.0%	99.8%	Tyrosinemia, type I, 276700
FECH	107.9	100.0%	99.6%	Protoporphyria, erythropoietic, 1, 177000
FH	126.0	95.9%	89.5%	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800
GALT	165.3	100.0%	100.0%	Galactosemia, 230400
GANAB	113.2	100.0%	98.9%	Polycystic kidney disease 3, 600666
GBA	180.2	100.0%	100.0%	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	152.5	100.0%	99.5%	Polyglucosan body disease, adult form, 263570 Glycogen storage disease IV, 232500

GFM1	104.4	100.0%	98.9%	Combined oxidative phosphorylation deficiency 1, 609060
GLIS3	133.1	100.0%	99.6%	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199
HADHA	74.6	96.1%	89.6%	LCHAD deficiency, 609016 HELLP syndrome, maternal, of pregnancy, 609016 Fatty liver, acute, of pregnancy, 609016 Trifunctional protein deficiency, 609015
HAMP	185.1	100.0%	100.0%	Hemochromatosis, type 2B, 613313
HFE	114.7	100.0%	99.1%	Hemochromatosis, 235200
HNF1B	130.8	99.8%	97.9%	Renal cysts and diabetes syndrome, 137920 Diabetes mellitus, noninsulin-dependent, 125853
HSD17B4	106.4	95.5%	93.1%	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
HSD3B7	157.0	99.8%	97.4%	Bile acid synthesis defect, congenital, 1, 607765
IARS	124.2	99.9%	99.2%	Growth retardation, impaired intellectual development, hypotonia, and hepatopathy, 617093
IFT140	127.6	100.0%	99.6%	Retinitis pigmentosa 80, 617781 Short-rib thoracic dysplasia 9 with or without polydactyly, 266920
IFT172	98.4	100.0%	99.5%	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT43	119.5	100.0%	100.0%	?Cranioectodermal dysplasia 3, 614099 Short-rib thoracic dysplasia 18 with polydactyly, 617866 ?Retinitis pigmentosa 81, 617871
INSR	123.5	99.4%	96.1%	Hyperinsulinemic hypoglycemia, familial, 5, 609968 Rabson-Mendenhall syndrome, 262190 Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549 Leprechaunism, 246200
INVS	147.7	100.0%	100.0%	Nephronophthisis 2, infantile, 602088
JAG1	143.4	99.4%	97.6%	Alagille syndrome 1, 118450 Tetralogy of Fallot, 187500 ?Deafness, congenital heart defects, and posterior embryotoxon, 617992
LARS	128.4	99.8%	98.0%	?Infantile liver failure syndrome 1, 615438
LRP5	183.1	99.9%	99.4%	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	106.2	99.9%	98.8%	Interstitial lung and liver disease, 615486 Charcot-Marie-Tooth disease, axonal, type 2U, 616280
MPV17	93.2	100.0%	98.5%	Charcot-Marie-Tooth disease, axonal, type 2EE, 618400 Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810
MTM1	78.4	98.4%	91.8%	Myotubular myopathy, X-linked, 310400
MYO5B	115.0	98.1%	94.9%	Microvillus inclusion disease, 251850
NBAS	138.4	99.9%	99.2%	Infantile liver failure syndrome 2, 616483 Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800
NHP2	135.0	100.0%	99.8%	Dyskeratosis congenita, autosomal recessive 2, 613987
NOP10	124.6	100.0%	100.0%	Dyskeratosis congenita, autosomal recessive 1, 224230
NOTCH2	130.5	100.0%	99.8%	Hajdu-Cheney syndrome, 102500 Alagille syndrome 2, 610205
NPC1	120.3	100.0%	99.4%	Niemann-Pick disease, type D, 257220 Niemann-Pick disease, type C1, 257220
NPC2	130.7	100.0%	99.9%	Niemann-pick disease, type C2, 607625
NPHP3	121.6	99.7%	98.3%	Meckel syndrome 7, 267010 Renal-hepatic-pancreatic dysplasia 1, 208540 Nephronophthisis 3, 604387
NR1H4	124.6	99.9%	97.5%	Cholestasis, progressive familial intrahepatic, 5, 617049
PEX1	126.3	100.0%	99.1%	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 Peroxisome biogenesis disorder 1A (Zellweger), 214100
PEX10	123.8	100.0%	98.4%	Peroxisome biogenesis disorder 6B, 614871 Peroxisome biogenesis disorder 6A (Zellweger), 614870
PEX12	125.4	100.0%	100.0%	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B, 266510
PEX13	189.6	100.0%	100.0%	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885
PEX14	144.7	99.8%	98.8%	Peroxisome biogenesis disorder 13A (Zellweger), 614887
PEX16	157.0	98.9%	95.7%	Peroxisome biogenesis disorder 8A (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877
PEX19	85.8	100.0%	98.9%	Peroxisome biogenesis disorder 12A (Zellweger), 614886
PEX2	137.4	100.0%	100.0%	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867
PEX26	105.1	100.0%	100.0%	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873

PEX3	108.6	100.0%	99.6%	Peroxisome biogenesis disorder 10A (Zellweger), 614882 ?Peroxisome biogenesis disorder 10B, 617370
PEX5	115.8	100.0%	99.4%	Peroxisome biogenesis disorder 2B, 202370 Rhizomelic chondrodysplasia punctata, type 5, 616716 Peroxisome biogenesis disorder 2A (Zellweger), 214110
PEX6	117.6	99.1%	93.9%	Peroxisome biogenesis disorder 4B, 614863 Heimler syndrome 2, 616617 Peroxisome biogenesis disorder 4A (Zellweger), 614862
PEX7	108.8	91.3%	91.0%	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100
PKD1	41.1	44.3%	36.9%	Polycystic kidney disease 1, 173900
PKD2	105.1	99.2%	96.8%	Polycystic kidney disease 2, 613095
PKHD1	132.5	100.0%	99.5%	Polycystic kidney disease 4, with or without hepatic disease, 263200
POLG	124.4	100.0%	99.8%	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	165.9	100.0%	100.0%	Obesity, adrenal insufficiency, and red hair due to POMC deficiency, 609734
PRKCSH	165.4	99.8%	96.3%	Polycystic liver disease 1, 174050
RFX6	150.2	100.0%	99.7%	Mitchell-Riley syndrome, 615710
RPGRIP1L	124.2	96.8%	95.8%	COACH syndrome, 216360 Meckel syndrome 5, 611561 Joubert syndrome 7, 611560
SC5D	149.4	100.0%	99.6%	Lathosterolosis, 607330
SCO1	105.2	100.0%	99.6%	Mitochondrial complex IV deficiency, 220110
SEC61B	112.6	97.6%	85.9%	No OMIM Disease ID
SEC63	76.2	87.1%	79.3%	Polycystic liver disease 2, 617004
SERPINA1	113.2	100.0%	99.8%	Hemorrhagic diathesis due to antithrombin Pittsburgh, 613490 Emphysema-cirrhosis, due to AAT deficiency, 613490 Emphysema due to AAT deficiency, 613490
SLC25A13	118.2	99.9%	98.1%	Citrullinemia, adult-onset type II, 603471 Citrullinemia, type II, neonatal-onset, 605814
SLC40A1	121.3	100.0%	99.9%	Hemochromatosis, type 4, 606069
SMPD1	161.8	100.0%	99.6%	Niemann-Pick disease, type A, 257200 Niemann-Pick disease, type B, 607616
STN1	82.8	100.0%	99.6%	Cerebroretinal microangiopathy with calcifications and cysts 2, 617341

TALDO1	158.9	100.0%	99.8%	Transaldolase deficiency, 606003
TERC	NC	NC	NC	Dyskeratosis congenita, autosomal dominant 1, 127550
TERT	160.1	99.9%	99.0%	No OMIM disease ID
TFR2	138.0	99.8%	98.7%	Hemochromatosis, type 3, 604250
TJP2	114.9	94.0%	93.6%	Hypercholanemia, familial, 607748 Cholestasis, progressive familial intrahepatic 4, 615878
TMEM67	80.6	99.3%	93.5%	Meckel syndrome 3, 607361 ?RHYNS syndrome, 602152 Nephronophthisis 11, 613550 COACH syndrome, 216360 Joubert syndrome 6, 610688
TRAF3IP1	87.5	99.1%	96.7%	Senior-Loken syndrome 9, 616629
TRMU	106.5	100.0%	99.5%	Liver failure, transient infantile, 613070
TTC37	131.5	99.9%	98.9%	Trichohepatoenteric syndrome 1, 222470
TWNK	170.3	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
UBR1	118.2	99.9%	98.9%	Johanson-Blizzard syndrome, 243800
UGT1A1	192.9	100.0%	100.0%	Hyperbilirubinemia, familial transient neonatal, 237900 Crigler-Najjar syndrome, type I, 218800 Crigler-Najjar syndrome, type II, 606785
VIPAS39	114.4	100.0%	99.9%	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VPS33B	111.7	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.
Median Coverage describes the average number of reads seen across 50 exomes.
% 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 Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : December 11th , 2019.
This list is accurate for panel version DG 2.17
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors