WES LIVER DISORDERS DG 3.5

Gene	Median coverage	% covered >10x	% covered >20x	OMIM disease ID
ABCB11	135.8	100.0	99.7	601847
ABCB4	146.8	100.0	100.0	602347
ABCC2	124.9	100.0	100.0	237500
ABCD3	148.5	100.0	100.0	616278
ACOX2	131.0	100.0	100.0	617308
ACTA2	147.4	99.9	99.1	613834
ACTG2	156.8	100.0	100.0	155310
ADK	130.8	90.9	90.9	614300
AHCY	141.9	100.0	100.0	613752
AKR1D1	140.1	100.0	100.0	235555
ALDOB	129.9	100.0	100.0	229600
ALG8	139.1	96.1	96.1	608104;617874
AMACR	152.1	100.0	100.0	214950
ANKS6	127.9	99.9	99.4	615382
AP1S1	114.6	100.0	100.0	609313
ATP7B	132.0	100.0	100.0	277900
ATP8B1	138.2	100.0	100.0	211600
BAAT	139.9	100.0	100.0	607748
BCS1L	133.1	100.0	100.0	124000
BLVRA	118.6	100.0	99.9	614156
CC2D2A	138.0	98.2	98.2	216360
CEP83	149.0	100.0	100.0	615862
CFC1	217.5	100.0	100.0	605376
CFTR	166.7	100.0	100.0	219700
CHD8	129.5	100.0	100.0	615032

CHRM3	126.9	100.0	100.0	100100
CHRNA3	134.6	100.0	100.0	191800
CLDN1	135.6	100.0	100.0	607626
CLMP	126.9	100.0	100.0	615237
COG7	127.2	100.0	100.0	608779
CYP27A1	136.9	100.0	100.0	213700
CYP7B1	158.7	100.0	100.0	613812
DCDC2	140.0	100.0	100.0	617394
DGUOK	135.8	100.0	100.0	251880
DHCR7	129.7	100.0	100.0	270400
DKC1	105.8	100.0	100.0	305000
DNAJB11	136.2	100.0	100.0	618061
EDNRB	148.0	100.0	100.0	600501
EPHX1	117.0	100.0	100.0	607748
ERBB3	123.4	100.0	100.0	243180
ETFDH	140.8	100.0	100.0	231680
FAH	128.4	100.0	100.0	276700
FECH	140.8	100.0	100.0	177000
FH	143.5	100.0	100.0	606812
FLNA	105.8	100.0	99.9	300048
GALT	130.4	100.0	100.0	230400
GANAB	124.2	100.0	100.0	600666
GBA	136.2	100.0	100.0	230800
GBE1	156.7	100.0	99.9	232500
GDNF	164.1	100.0	100.0	209880
GFM1	151.0	100.0	100.0	609060
GLI3	127.4	100.0	100.0	146510
GLIS3	137.8	100.0	100.0	610199
HADHA	128.3	100.0	100.0	609016
HAMP	115.9	100.0	100.0	613313

HFE	152.9	100.0	100.0	235200
HNF1B	131.1	100.0	100.0	137920
HSD17B4	142.8	96.6	96.6	261515
HSD3B7	128.2	100.0	100.0	607765
IARS1	139.6	100.0	100.0	617093
IFT140	135.5	100.0	100.0	266920
IFT172	131.1	100.0	100.0	615630
IFT43	133.8	100.0	100.0	617866
INSR	117.2	100.0	100.0	246200;609968
INVS	121.7	100.0	100.0	602088
JAG1	137.5	100.0	100.0	118450
KIF12	133.9	100.0	100.0	619662
LARS1	144.1	100.0	100.0	615438
LMOD1	119.6	100.0	100.0	619362
LRP5	121.4	100.0	100.0	617875
MARS1	128.4	100.0	100.0	615486
MPV17	140.8	100.0	100.0	256810
MTM1	108.2	99.7	99.2	310400
MYH11	129.0	100.0	100.0	619351
MYL9	119.2	100.0	100.0	619365
MYLK	127.5	100.0	100.0	249210
MYO5B	123.7	100.0	99.9	251850
NBAS	144.1	100.0	99.9	616483
NHP2	118.6	100.0	100.0	613987
NOP10	131.5	100.0	100.0	224230
NOTCH2	149.2	100.0	100.0	610205
NPC1	133.9	100.0	100.0	257220
NPC2	136.1	100.0	100.0	607625
NPHP3	145.9	100.0	100.0	208540
NR1H4	144.8	100.0	100.0	617049

PEX1	145.2	100.0	100.0	601539;214100
PEX10	124.7	100.0	100.0	614870
PEX12	132.3	100.0	100.0	614859
PEX13	151.6	100.0	100.0	614883
PEX14	128.8	100.0	100.0	614887
PEX16	133.1	100.0	100.0	614876
PEX19	128.7	100.0	100.0	614886
PEX2	165.5	100.0	100.0	614866
PEX26	126.8	100.0	100.0	614872
PEX3	153.8	100.0	100.0	614882
PEX5	132.0	100.0	100.0	214110
PEX6	133.9	100.0	100.0	614862
PEX7	139.0	91.2	91.2	215100
PKD1	156.6	99.9	99.7	173900
PKD2	139.7	100.0	100.0	613095
PKHD1	135.3	100.0	100.0	263200
POLG	138.3	100.0	100.0	203700
POMC	149.8	100.0	100.0	609734
PRKCSH	126.6	100.0	100.0	174050
RAD21	145.4	100.0	100.0	611376
RFX6	142.4	100.0	100.0	615710
RINT1	145.8	100.0	100.0	-
RPGRIP1L	147.0	100.0	100.0	216360
SC5D	149.4	100.0	100.0	607330
SCO1	154.5	100.0	100.0	220110
SCYL1	121.8	100.0	100.0	No OMIM phenotype
SEC61B	144.6	100.0	100.0	617004
SEC63	143.7	100.0	100.0	617004
SEMA7A	125.7	100.0	100.0	619874
SERPINA1	138.2	100.0	100.0	613490

SGO1	155.3	100.0	100.0	616201
SLC25A13	144.4	100.0	100.0	605814
SLC40A1	146.4	100.0	100.0	606069
SLC51A	125.9	100.0	100.0	619484
SMPD1	139.2	100.0	100.0	257200;607616
SOX10	142.9	100.0	100.0	609136
STN1	133.5	100.0	100.0	617341
TALDO1	122.3	100.0	100.0	606003
TERC				127550
TERT	156.5	100.0	100.0	613989
TFR2	117.3	100.0	100.0	604250
TJP2	144.2	100.0	100.0	615878
TMEM67	147.4	99.5	97.5	216360
TRAF3IP1	134.9	100.0	100.0	616629
TRMU	124.4	100.0	100.0	613070
TTC37	151.1	100.0	100.0	222470
TWNK	117.5	100.0	100.0	271245
TYMP	175.3	100.0	100.0	603041
UBR1	140.0	98.0	98.0	243800
UGT1A1	150.4	100.0	100.0	218800;606785
USP53	152.3	100.0	100.0	619658
VIPAS39	129.6	100.0	100.0	613404
VPS33B	128.9	100.0	100.0	208085
ZFYVE19	123.7	100.0	100.0	619849

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 default chemistry for all WES samples. Agilent V5 was the default chemistry until Q3 2021.
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 no value for coverage 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 1st, 2021.

Ad 1. “No OMIM phenotype” signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors