

WES LIVER DISORDERS DG 3.1

<i>Gene</i>	<i>Median coverage</i>	<i>% covered >10x</i>	<i>% covered >20x</i>	<i>OMIM disease ID</i>
ABCB11	170.4	100.0	99.7	601847
ABCB4	151.8	99.9	99.6	602347
ABCC2	134.7	100.0	99.9	237500
ABCD3	125.8	99.8	97.7	616278
ACOX2	120.6	100.0	99.2	617308
ACTA2	107.2	100.0	99.0	613834
ACTG2	119.3	99.9	98.2	155310
ADK	96.9	84.1	81.0	614300
AHCY	117.4	100.0	99.2	613752
AKR1D1	117.4	100.0	99.4	235555
ALDOB	152.6	99.4	96.6	229600
ALG8	132.5	97.2	95.6	608104;617874
AMACR	159.5	100.0	100.0	214950
ANKS6	95.9	93.8	89.5	615382
AP1S1	124.8	99.9	99.5	609313
ATP7B	146.0	99.9	99.2	277900
ATP8B1	141.0	96.5	94.0	211600
BAAT	134.8	99.8	98.4	607748
BCS1L	158.0	100.0	100.0	124000
BLVRA	120.4	100.0	99.4	614156
CC2D2A	131.6	98.5	96.5	216360
CEP83	133.1	99.8	97.4	615862
CFC1	122.7	84.2	74.1	605376
CFTR	140.6	99.6	97.9	219700
CHD8	151.5	100.0	99.9	615032

CHRM3	144.2	100.0	100.0	100100
CHRNA3	137.9	100.0	99.4	191800
CLDN1	132.0	100.0	100.0	607626
CLMP	95.9	100.0	99.6	615237
COG7	125.0	100.0	100.0	608779
CYP27A1	180.7	98.9	96.7	213700
CYP7B1	131.5	98.0	92.8	613812
DCDC2	174.2	100.0	99.9	617394
DGUOK	124.5	100.0	99.4	251880
DHCR7	152.1	100.0	100.0	270400
DKC1	108.6	99.8	98.7	305000
DNAJB11	128.6	100.0	99.5	618061
EDNRB	134.4	98.0	93.8	600501
EPHX1	137.1	99.9	98.8	607748
ETFDH	141.8	100.0	99.8	231680
FAH	135.8	100.0	100.0	276700
FECH	120.3	100.0	100.0	177000
FH	147.0	92.1	88.3	606812
FLNA	138.5	100.0	99.9	300048
GALT	167.6	100.0	99.7	230400
GANAB	129.4	99.9	99.0	600666
GBA	202.3	100.0	100.0	230800
GBE1	200.1	100.0	99.6	232500
GDNF	204.6	100.0	100.0	209880
GFM1	131.2	99.9	99.4	609060
GLI3	140.8	98.5	98.0	146510
GLIS3	136.1	98.6	98.2	610199
HADHA	87.5	97.2	91.6	609016
HAMP	192.7	100.0	100.0	613313
HFE	127.9	100.0	99.7	235200

HNF1B	133.8	99.3	96.1	137920
HSD17B4	130.0	95.4	93.1	261515
HSD3B7	142.9	99.1	95.5	607765
IARS1	150.2	100.0	99.6	617093
IFT140	124.5	99.8	98.8	266920
IFT172	107.8	99.9	99.1	615630
IFT43	134.3	100.0	100.0	617866
INSR	138.2	97.8	94.7	246200;609968
INVS	163.7	100.0	100.0	602088
JAG1	147.4	97.7	96.8	118450
LARS1	164.6	99.8	98.4	615438
LRP5	179.9	98.5	98.1	617875
MARS1	114.4	99.7	97.4	615486
MPV17	97.4	100.0	97.2	256810
MTM1	95.6	99.0	93.3	310400
MYO5B	125.2	99.1	96.2	251850
NBAS	169.2	100.0	99.6	616483
NHP2	146.1	100.0	100.0	613987
NOP10	147.4	100.0	99.8	224230
NOTCH2	146.7	100.0	99.5	610205
NPC1	136.4	99.6	98.7	257220
NPC2	160.5	100.0	99.6	607625
NPHP3	143.4	99.7	98.4	208540
NR1H4	147.9	99.8	98.5	617049
PEX1	155.5	99.9	99.4	601539;214100
PEX10	101.8	96.8	89.7	614870
PEX12	150.7	100.0	100.0	614859
PEX13	208.9	100.0	100.0	614883
PEX14	132.7	96.7	90.8	614887
PEX16	158.6	97.9	94.2	614876

PEX19	103.7	99.9	98.5	614886
PEX2	158.8	100.0	100.0	614866
PEX26	94.0	100.0	100.0	614872
PEX3	125.7	100.0	99.3	614882
PEX5	124.0	99.9	99.0	214110
PEX6	113.0	94.5	86.7	614862
PEX7	135.9	87.8	80.7	215100
PKD1	27.3	39.2	30.0	173900
PKD2	108.4	95.5	91.1	613095
PKHD1	154.2	100.0	99.6	263200
POLG	111.5	100.0	99.3	203700
POMC	114.0	100.0	100.0	609734
PRKCSH	160.1	99.8	95.4	174050
RAD21	103.0	99.2	96.6	611376
RFX6	177.2	100.0	99.6	615710
RPGRIP1L	155.5	96.7	95.7	216360
SC5D	192.4	100.0	99.5	607330
SCO1	115.4	97.1	93.8	220110
SEC61B	118.5	99.1	92.4	617004
SEC63	99.2	91.2	83.3	617004
SERPINA1	110.7	100.0	100.0	613490
SGO1	137.6	99.9	98.9	616201
SLC25A13	146.4	100.0	99.7	605814
SLC40A1	134.9	100.0	99.5	606069
SMPD1	171.4	100.0	100.0	257200;607616
STN1	102.9	100.0	100.0	617341
TALDO1	151.8	100.0	97.9	606003
TERC				127550
TERT	132.4	96.2	94.5	613989
TFR2	120.0	99.1	97.8	604250

TJP2	117.6	92.8	92.5	615878
TMEM67	96.9	99.5	95.0	216360
TRAF3IP1	91.1	99.6	97.6	616629
TRMU	103.5	100.0	100.0	613070
TTC37	165.8	100.0	99.3	222470
TWNK	202.8	100.0	100.0	271245
TYMP	97.1	100.0	97.0	603041
UBR1	143.8	99.9	99.1	243800
UGT1A1	227.3	100.0	100.0	218800;606785
VIPAS39	133.8	100.0	100.0	613404
VPS33B	128.1	100.0	100.0	208085

Gene symbols used follow HGNC 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 : October 1st, 2016.

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