# WES LIVER DISORDERS DG 3.3

| Gene | Median coverage | % covered >10x | % covered >20x | OMIM disease ID |
|---|---|---|---|---|
| ABCB11 | 174.7 | 100.0 | 100.0 | 601847 |
| ABCB4 | 167.3 | 100.0 | 100.0 | 602347 |
| ABCC2 | 156.2 | 100.0 | 100.0 | 237500 |
| ABCD3 | 139.1 | 100.0 | 100.0 | 616278 |
| ACOX2 | 161.5 | 100.0 | 100.0 | 617308 |
| ACTA2 | 168.2 | 100.0 | 100.0 | 613834 |
| ACTG2 | 186.9 | 100.0 | 100.0 | 155310 |
| ADK | 114.4 | 84.5 | 84.5 | 614300 |
| AHCY | 167.4 | 100.0 | 100.0 | 613752 |
| AKR1D1 | 146.7 | 100.0 | 100.0 | 235555 |
| ALDOB | 147.3 | 100.0 | 100.0 | 229600 |
| ALG8 | 147.6 | 96.6 | 96.6 | 608104;617874 |
| AMACR | 192.5 | 100.0 | 100.0 | 214950 |
| ANKS6 | 172.9 | 99.9 | 99.1 | 615382 |
| AP1S1 | 134.4 | 100.0 | 100.0 | 609313 |
| ATP7B | 173.5 | 100.0 | 100.0 | 277900 |
| ATP8B1 | 152.9 | 100.0 | 100.0 | 211600 |
| BAAT | 240.7 | 100.0 | 100.0 | 607748 |
| BCS1L | 173.9 | 100.0 | 100.0 | 124000 |
| BLVRA | 146.3 | 100.0 | 100.0 | 614156 |
| CC2D2A | 149.2 | 97.1 | 97.1 | 216360 |
| CEP83 | 133.8 | 100.0 | 100.0 | 615862 |
| CFC1 | 274.6 | 100.0 | 100.0 | 605376 |
| CFTR | 150.7 | 100.0 | 100.0 | 219700 |
| CHD8 | 169.6 | 100.0 | 100.0 | 615032 |

| | | | | |
|---|---|---|---|---|
| CHRM3 | 186.6 | 100.0 | 100.0 | 100100 |
| CHRNA3 | 183.7 | 100.0 | 100.0 | 191800 |
| CLDN1 | 173.7 | 100.0 | 100.0 | 607626 |
| CLMP | 175.7 | 100.0 | 100.0 | 615237 |
| COG7 | 154.7 | 100.0 | 100.0 | 608779 |
| CYP27A1 | 173.9 | 100.0 | 100.0 | 213700 |
| CYP7B1 | 142.1 | 100.0 | 100.0 | 613812 |
| DCDC2 | 165.1 | 100.0 | 100.0 | 617394 |
| DGUOK | 144.7 | 100.0 | 100.0 | 251880 |
| DHCR7 | 173.2 | 100.0 | 100.0 | 270400 |
| DKC1 | 139.9 | 100.0 | 100.0 | 305000 |
| DNAJB11 | 138.2 | 100.0 | 100.0 | 618061 |
| EDNRB | 168.2 | 100.0 | 100.0 | 600501 |
| EPHX1 | 170.8 | 100.0 | 100.0 | 607748 |
| ETFDH | 148.8 | 100.0 | 100.0 | 231680 |
| FAH | 144.4 | 100.0 | 100.0 | 276700 |
| FECH | 155.0 | 100.0 | 100.0 | 177000 |
| FH | 185.4 | 100.0 | 100.0 | 606812 |
| FLNA | 204.7 | 100.0 | 100.0 | 300048 |
| GALT | 171.0 | 100.0 | 100.0 | 230400 |
| GANAB | 160.1 | 100.0 | 100.0 | 600666 |
| GBA | 218.8 | 100.0 | 100.0 | 230800 |
| GBE1 | 152.1 | 100.0 | 100.0 | 232500 |
| GDNF | 193.4 | 100.0 | 100.0 | 209880 |
| GFM1 | 149.0 | 100.0 | 100.0 | 609060 |
| GLI3 | 193.7 | 100.0 | 100.0 | 146510 |
| GLIS3 | 195.5 | 100.0 | 100.0 | 610199 |
| HADHA | 163.0 | 100.0 | 100.0 | 609016 |
| HAMP | 158.4 | 100.0 | 100.0 | 613313 |
| HFE | 176.9 | 100.0 | 100.0 | 235200 |

| | | | | |
|---|---|---|---|---|
| HNF1B | 234.4 | 100.0 | 100.0 | 137920 |
| HSD17B4 | 133.6 | 96.6 | 96.6 | 261515 |
| HSD3B7 | 187.4 | 100.0 | 100.0 | 607765 |
| IARS1 | 145.1 | 100.0 | 100.0 | 617093 |
| IFT140 | 186.7 | 100.0 | 100.0 | 266920 |
| IFT172 | 154.8 | 100.0 | 100.0 | 615630 |
| IFT43 | 145.7 | 100.0 | 100.0 | 617866 |
| INSR | 157.4 | 100.0 | 100.0 | 246200;609968 |
| INVS | 168.3 | 100.0 | 100.0 | 602088 |
| JAG1 | 180.2 | 100.0 | 100.0 | 118450 |
| LARS1 | 147.7 | 100.0 | 100.0 | 615438 |
| LRP5 | 223.0 | 100.0 | 100.0 | 617875 |
| MARS1 | 161.5 | 100.0 | 100.0 | 615486 |
| MPV17 | 157.0 | 100.0 | 100.0 | 256810 |
| MTM1 | 145.2 | 100.0 | 100.0 | 310400 |
| MYO5B | 148.2 | 100.0 | 100.0 | 251850 |
| NBAS | 161.1 | 100.0 | 100.0 | 616483 |
| NHP2 | 158.2 | 100.0 | 100.0 | 613987 |
| NOP10 | 150.8 | 100.0 | 100.0 | 224230 |
| NOTCH2 | 247.6 | 100.0 | 100.0 | 610205 |
| NPC1 | 157.1 | 100.0 | 100.0 | 257220 |
| NPC2 | 143.9 | 100.0 | 100.0 | 607625 |
| NPHP3 | 147.4 | 100.0 | 100.0 | 208540 |
| NR1H4 | 162.2 | 100.0 | 100.0 | 617049 |
| PEX1 | 148.7 | 100.0 | 100.0 | 601539;214100 |
| PEX10 | 201.3 | 100.0 | 100.0 | 614870 |
| PEX12 | 159.1 | 100.0 | 100.0 | 614859 |
| PEX13 | 172.8 | 100.0 | 100.0 | 614883 |
| PEX14 | 167.4 | 100.0 | 100.0 | 614887 |
| PEX16 | 183.1 | 100.0 | 100.0 | 614876 |

| | | | | |
|---|---|---|---|---|
| PEX19 | 152.9 | 100.0 | 100.0 | 614886 |
| PEX2 | 189.3 | 100.0 | 100.0 | 614866 |
| PEX26 | 158.3 | 100.0 | 100.0 | 614872 |
| PEX3 | 130.0 | 100.0 | 100.0 | 614882 |
| PEX5 | 170.9 | 100.0 | 100.0 | 214110 |
| PEX6 | 189.6 | 100.0 | 100.0 | 614862 |
| PEX7 | 137.2 | 91.3 | 91.3 | 215100 |
| PKD1 | 228.8 | 99.8 | 99.6 | 173900 |
| PKD2 | 158.5 | 100.0 | 100.0 | 613095 |
| PKHD1 | 170.4 | 100.0 | 100.0 | 263200 |
| POLG | 180.5 | 100.0 | 100.0 | 203700 |
| POMC | 199.9 | 100.0 | 100.0 | 609734 |
| PRKCSH | 169.5 | 100.0 | 100.0 | 174050 |
| RAD21 | 171.5 | 100.0 | 100.0 | 611376 |
| RFX6 | 154.6 | 100.0 | 100.0 | 615710 |
| RINT1 | 154.1 | 100.0 | 100.0 | - |
| RPGRIP1L | 146.2 | 99.9 | 99.4 | 216360 |
| SC5D | 141.5 | 100.0 | 100.0 | 607330 |
| SCO1 | 196.9 | 100.0 | 100.0 | 220110 |
| SCYL1 | 170.8 | 100.0 | 100.0 | No OMIM phenotype |
| SEC61B | 170.5 | 100.0 | 100.0 | 617004 |
| SEC63 | 144.7 | 100.0 | 100.0 | 617004 |
| SERPINA1 | 245.5 | 100.0 | 100.0 | 613490 |
| SGO1 | 152.1 | 100.0 | 100.0 | 616201 |
| SLC25A13 | 152.7 | 100.0 | 100.0 | 605814 |
| SLC40A1 | 169.6 | 100.0 | 100.0 | 606069 |
| SMPD1 | 209.7 | 100.0 | 100.0 | 257200;607616 |
| STN1 | 141.9 | 100.0 | 100.0 | 617341 |
| TALDO1 | 160.4 | 100.0 | 100.0 | 606003 |
| TERC | | | | 127550 |

| | | | | |
|---|---|---|---|---|
| TERT | 267.9 | 100.0 | 100.0 | 613989 |
| TFR2 | 183.4 | 100.0 | 100.0 | 604250 |
| TJP2 | 176.4 | 98.8 | 98.8 | 615878 |
| TMEM67 | 127.3 | 100.0 | 99.9 | 216360 |
| TRAF3IP1 | 161.1 | 100.0 | 100.0 | 616629 |
| TRMU | 161.4 | 100.0 | 100.0 | 613070 |
| TTC37 | 146.6 | 100.0 | 100.0 | 222470 |
| TWNK | 185.1 | 100.0 | 100.0 | 271245 |
| TYMP | 204.3 | 100.0 | 100.0 | 603041 |
| UBR1 | 139.0 | 98.0 | 98.0 | 243800 |
| UGT1A1 | 203.8 | 100.0 | 100.0 | 218800;606785 |
| VIPAS39 | 132.5 | 100.0 | 100.0 | 613404 |
| VPS33B | 140.0 | 100.0 | 100.0 | 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.*
*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.*