MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Cholestasis Gene Panel

Next-generation sequencing (NGS) and/or Sanger sequencing is performed to test for the presence of variants in coding regions and intron/exon boundaries of the genes analyzed. NGS and/or a polymerase chain reaction (PCR)-based quantitative method is performed to test for the presence of deletions and duplications in the genes analyzed. Confirmation of select reportable variants may be performed by alternate methodologies based on internal laboratory criteria.

Genomic Build: GRCh37 (hg19) unless otherwise specified

Gene	Reference Transcript[a]
ABCB11	NM_003742.4
ABCB4	NM_000443.4
ABCC2	NM_000392.5
ABCG5	NM_022436.3
ABCG8	NM_022437.3
ABHD5	NM_016006.6
ACOX1	NM_004035.7
AGL	NM_000642.3
AGPAT2	NM_006412.4
AKR1D1	NM_005989.4
ALDOA	NM_000034.3
ALDOB	NM_000035.4
AMACR	NM_014324.6
ARSB	NM_000046.5
ASAH1[b]	NM_177924.5
ATP8B1	NM_005603.6
BAAT	NM_001701.4
BSCL2	NM_032667.6
CAVIN1	NM_012232.6
CC2D2A	NM_001080522.2
CFTR[b]	NM_000492.4
CIDEC	NM_001199623.1
CLDN1	NM_021101.5
CYP27A1	NM_000784.4
CYP7A1	NM_000780.4
CYP7B1	NM_004820.5
DCDC2	NM_016356.5
DGUOK	NM_080916.3
DHCR7	NM_001360.2

Gene	Reference Transcript[a]
EHHADH	NM_001966.4
FAH	NM_000137.3
FBP1	NM_000507.4
FUCA1	NM_000147.4
G6PC	NM_000151.4
GAA	NM_000152.5
GALNS	NM_000512.5
GBA	NM_000157.4
GBE1	NM_000158.4
GLB1	NM_000404.4
GNE	NM_001128227.3
GNPTAB[b]	NM_024312.5
GNS[b]	NM_002076.4
GUSB	NM_000181.4
HADHA[b]	NM_000182.5
HGSNAT	NM_152419.3
HNF1B	NM_000458.4
HSD17B4	NM_001199291.3
HSD3B7	NM_025193.4
IDS	NM_000202.8
IDUA	NM_000203.5
INVS	NM_014425.5
JAG1	NM_000214.3
KCNH1	NM_172362.3
LIPA	NM_000235.4
MAN2B1	NM_000528.4
MKS1	NM_017777.4
MPV17	NM_002437.5
MVK	NM_000431.4

Targeted Genes and Methodology Details for Cholestasis Gene Panel (continued)

Gene	Reference Transcript[a]
NAGLU	NM_000263.4
NEU1	NM_000434.4
NOTCH2[b,c]	NM_024408.4
NPC1[b]	NM_000271.5
NPC2	NM_006432.4
NPHP1[b]	NM_000272.4
NPHP3[b]	NM_153240.5
NPHP4	NM_015102.5
NR1H4[b]	NM_005123.4
PEPD	NM_000285.4
PEX1	NM_000466.3
PEX10	NM_153818.1
PEX11B	NM_003846.3
PEX12	NM_000286.3
PEX13	NM_002618.4
PEX14	NM_004565.3
PEX16	NM_057174.2
PEX19	NM_002857.3
PEX2	NM_000318.3
PEX26	NM_017929.6
PEX3	NM_003630.3
PEX5	NM_001131023.1
PEX6	NM_000287.4
PEX7	NM_000288.4
PHKA2[b]	NM_000292.3
PHKB[b]	NM_000293.3
PHKG2	NM_000294.3

Gene	Reference Transcript[a]
PKHD1	NM_138694.4
PNPLA2	NM_020376.4
POLG	NM_002693.2
PRKAG2[b]	NM_016203.4
PSAP	NM_002778.4
PYGL	NM_002863.5
SCP2[b]	NM_002979.5
SERPINA1	NM_000295.5
SGSH	NM_000199.5
SLC10A1	NM_003049.4
SLC10A2	NM_000452.3
SLC17A5	NM_012434.5
SLC25A13	NM_014251.3
SLC27A5	NM_012254.3
SLC37A4	ENST00000545985.1
SLC7A7	NM_001126106.2
SMPD1	NM_000543.5
SUMF1	NM_182760.4
TALDO1	NM_006755.2
TJP2	NM_004817.4
TMEM216	NM_001173990.3
TRIM37	NM_015294.6
TRMU	NM_018006.5
UGT1A1	NM_000463.3
VIPAS39	NM_022067.4
VPS33A	NM_022916.6
VPS33B	NM_018668.4

[a] Reference transcript numbers may be updated due to transcript re-versioning. Always refer to the final patient report for gene transcript information referenced at the time of testing.

[b] There are regions of this gene that cannot be effectively analyzed for the presence of copy number variants.

[c] There are regions of this gene that cannot be effectively amplified for sequencing as a result of technical limitations of the assay, including regions of homology, high GC content, and repetitive sequences.

To verify if a specific region/exon/variant is covered by this assay, contact a laboratory genetic counselor at 800-533-1710.