MAYO CLINIC LABORATORIES | *Targeted Genes and Methodology Details for Cholestasis Gene Panel*

The following applies to CHLGP / Cholestasis Gene Panel. Testing is performed to evaluate for the presence of variants in coding regions and extending to +/- 30 base pairs of adjacent intronic sequence on either side of the coding exons of the genes analyzed. In addition, the analysis will cover select non-coding variants. Next-generation sequencing and/or a polymerase chain reaction-based quantitative method is performed to test for the presence of copy number variants in the genes analyzed. Confirmation of select reportable variants may be performed by alternate methodologies based on internal laboratory criteria.

This list is current from February 2021 to the present. This document is intended to highlight additional evaluations for variants of high clinical interest to providers as well as technical limitations. However, this document does not comprehensively reflect all genomic regions covered by this test. For questions regarding transcripts, genes or regions covered, contact the laboratory at 800-533-1710.

Genomic Build: GRCh37 (hg19) unless otherwise specified

Gene	Reference Transcript[a]	Gene	Reference Transcript[a]
ABCB11	NM_003742.4	DCDC2	NM_016356.5
ABCB4	NM_000443.4	DGUOK	NM_080916.3
ABCC2	NM_000392.5	DHCR7	NM_001360.2
ABCG5	NM_022436.3	EHHADH	NM_001966.4
ABCG8	NM_022437.3	FAH	NM_000137.3
ABHD5	NM_016006.6	FBP1	NM_000507.4
ACOX1	NM_004035.7	FUCA1	NM_000147.4
AGL	NM_000642.3	G6PC	NM_000151.4
AGPAT2	NM_006412.4	GAA	NM_000152.5
AKR1D1	NM_005989.4	GALNS	NM_000512.5
ALDOA	NM_000034.3	GBA	NM_000157.4
ALDOB	NM_000035.4	GBE1	NM_000158.4
AMACR	NM_014324.6	GLB1	NM_000404.4
ARSB	NM_000046.5	GNE	NM_001128227.3
ASAH1[b]	NM_177924.5	GNPTAB[b]	NM_024312.5
ATP8B1	NM_005603.6	GNS[b]	NM_002076.4
BAAT	NM_001701.4	GUSB	NM_000181.4
BSCL2	NM_032667.6	HADHA[b]	NM_000182.5
CAVIN1	NM_012232.6	HGSNAT	NM_152419.3
CC2D2A	NM_001080522.2	HNF1B	NM_000458.4
CFTR[b]	NM_000492.4	HSD17B4	NM_001199291.3
CIDEC	NM_001199623.1	HSD3B7	NM_025193.4
CLDN1	NM_021101.5	IDS	NM_000202.8
CYP27A1	NM_000784.4	IDUA	NM_000203.5
CYP7A1	NM_000780.4	INVS	NM_014425.5
CYP7B1	NM_004820.5	JAG1	NM_000214.3

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Gene	Reference Transcript[a]	Gene	Reference Transcript[a]
KCNH1	NM_172362.3	PHKA2[b]	NM_000292.3
LIPA	NM_000235.4	PHKB[b]	NM_000293.3
MAN2B1	NM_000528.4	PHKG2	NM_000294.3
MKS1	NM_017777.4	PKHD1	NM_138694.4
MPV17	NM_002437.5	PNPLA2	NM_020376.4
MVK	NM_000431.4	POLG	NM_002693.2
NAGLU	NM_000263.4	PRKAG2[b]	NM_016203.4
NEU1	NM_000434.4	PSAP	NM_002778.4
NOTCH2[b,c]	NM_024408.4	PYGL	NM_002863.5
NPC1[b]	NM_000271.5	SCP2[b]	NM_002979.5
NPC2	NM_006432.4	SERPINA1	NM_000295.5
NPHP1[b]	NM_000272.4	SGSH	NM_000199.5
NPHP3[b]	NM_153240.5	SLC10A1	NM_003049.4
NPHP4	NM_015102.5	SLC10A2	NM_000452.3
NR1H4[b]	NM_005123.4	SLC17A5	NM_012434.5
PEPD	NM_000285.4	SLC25A13	NM_014251.3
PEX1	NM_000466.3	SLC27A5	NM_012254.3
PEX10	NM_153818.1	SLC37A4	ENST00000545985.1
PEX11B	NM_003846.3	SLC7A7	NM_001126106.2
PEX12	NM_000286.3	SMPD1	NM_000543.5
PEX13	NM_002618.4	SUMF1	NM_182760.4
PEX14	NM_004565.3	TALDO1	NM_006755.2
PEX16	NM_057174.2	TJP2	NM_004817.4
PEX19	NM_002857.3	TMEM216	NM_001173990.3
PEX2	NM_000318.3	TRIM37	NM_015294.6
PEX26	NM_017929.6	TRMU	NM_018006.5
PEX3	NM_003630.3	UGT1A1	NM_000463.3
PEX5	NM_001131023.1	VIPAS39	NM_022067.4
PEX6	NM_000287.4	VPS33A	NM_022916.6
PEX7	NM_000288.4	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.

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