

## **Cholestasis génpanel v:2025-05-15**

Panel tartalma 99 gén:

ABCB11, ABCB4, ABCC2, ADK, AKR1D1, ALDOB, AMACR, ATP7B, ATP8B1, BAAT, BCS1L, CC2D2A, CFTR, CLDN1, COG7, CREB3L3, CYP27A1, CYP7B1, DCDC2, DGUOK, EPCAM, FAH, FARSA, FARSB, GALE, GALK1, GALM, GALT, GBA, GBE1, GNAS, HADHA, HAMP, HFE, HJV, HNF1B, HSD3B7, IARS1, IARS1, INVS, JAG1, KIF12, LCT, LIPA, LMF1, LSR, MKS1, MMP15, MPI, MPV17, MVK, MYO5B, NBAS, NEUROG3, NOTCH2, NPC1, NPC2, NPHP1, NPHP3, NPHP4, NR1H4, PEX1, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PKHD1, POLG, PSKH1, RINT1, SCYL1, SERPINA1, SLC25A13, SLC26A3, SLCO1B1, SLCO1B3, SMPD1, SPINT2, TALDO1, TFR2, TJP2, TMEM216, TRMU, TTC37, UGT1A1, UNC45A, USP53, VIPAS39, VPS33B, VPS50, YARS1, ZFYVE19,

Forrás:

Cholestasis Panel

<https://blueprintgenetics.com/tests/panels/gastroenterology/cholestasis-panel/>

Cholestasis (Version 3.7)

<https://panelapp.genomicsengland.co.uk/panels/544/>

Neonatal cholestasis (Version 1.27)

<https://panelapp.genomicsengland.co.uk/panels/385/>

(Megtekintve: 2025-05-15)