

## **Genpanel PEDHEP (Pediatric Hepatology), version 23 – 170 gener**

Gener kopplade till medfödda leversjukdomar hos barn eller andra monogena sjukdomar med liknande klinisk bild

*ABCA1, ABCB11, ABCB4, ABCC2, ABCD3, ACOX2, ADK, AGL, AKR1D1, ALAS2, ALDOB, ALG3, ALG6, ALG8, ALG9, ALMS1, AMACR, ANKS6, ARG1, ASAH1, ASS1, ATP6AP1, ATP7B, ATP8B1, BAAT, BBS1, BCSIL, CC2D2A, CCDC115, CEP164, CEP290, CEP41, CEP83, CFTR, CLDN1, COG2, COG4, COG5, COG6, COG7, CSPP1, CYP27A1, CYP7A1, CYP7B1, DCDC2, DGUOK, DHCR7, DLD, DNAJB11, DNAJC21, DYNC2H1, DYNC2H2, DYNC2L11, EFL1, EIF2AK3, FAH, FBP1, FECH, FOCAD, G6PC, G6PD, GALE, GALT, GANAB, GATA6, GBE1, GDF1, GPD1, GPI, GUCY2D, GYS2, HAMP, HFE, HJV, HNF1B, HSD17B4, HSD3B7, IFT140, IFT172, IFT43, INPP5E, INVS, JAG1, KIF12, KRT18, KRT8, LARS1, LIPA, LRP5, LSR, MARS1, MMP15, MPI, MPV17, MTTP, MYO5B, NBAS, NEK1, NGLY1, NOTCH2, NPC1, NPC2, NPHP3, NRIH4, OFD1, PGM1, PHKA2, PHKB, PHKG2, PKD1, PKD2, PKHD1, PKLR, PMM2, POLG, PPM1F, PRF1, PRKCSH, PSKH1, PYGL, RINT1, RPGRIP1L, SBDS, SC5D, SCYL1, SEC61B, SEC63, SEMA7A, SERPINA1, SKIV2L, SLC10A1, SLC10A2, SLC25A13, SLC25A20, SLC27A5, SLC2A2, SLC37A4, SLC40A1, SLC51A, SLC51B, SLCO1B1, SLCO1B3, SMPD1, SRP54, TALDO1, TCTN2, TFAM, TFR2, TJP2, TMEM107, TMEM199, TMEM216, TMEM67, TRAF3IP1, TRMU, TTC26, TTC37, TULP3, TWNK, UGT1A1, UNC13D, UNC45A, USP53, VIPAS39, VPS33B, WDR19, WDR35, WDR83OS, XPNPEP3, ZFYVE19.*