

Genpanel PEDHEP (Pediatric Hepatology), version 22 – 172 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, ASAHI, ASS1, ATP6AP1, ATP7B, ATP8B1, BAAT, BBS1, BCS1L, CC2D2A, CCDC115, CEP164, CEP290, CEP41, CEP83, CFTR, CLDN1, COG2, COG4, COG5, COG6, COG7, CSPP1, CYP27A1, CYP7A1, CYP7B1, DCDC2, DGUOK, DHCR7, DNAJB11, DNAJC21, DYNC2H1, DYNC2I1, DYNC2I2, DYNC2L11, EFL1, EIF2AK3, FAH, FBPI, FECH, G6PC, G6PD, GALE, GALT, GANAB, GATA6, GBE1, GDF1, GPD1, GPI, GUCY2D, GYS2, HAMP, HFE, HJV, HNF1B, HSD17B4, HSD3B7, IFT140, IFT172, IFT43, IFT52, IFT81, INPP5E, INTU, INVS, JAG1, KIF12, KRT18, KRT8, LARS1, LIPA, LRP5, LSR, MARS1, MMP15, MPI, MPV17, MTP, MYO5B, NBAS, NEK1, NGLY1, NOTCH2, NPC1, NPC2, NPHP3, NR1H4, OFD1, PGM1, PHKA2, PHKB, PHKG2, PKD1, PKD2, PKHD1, PKLR, PLEC, PMM2, POLG, PPM1F, PRF1, PRKCSH, 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.