KAROLINSKA UNIVERSITY HOSPITAL

Centrum för Medfödda Metabola Sjukdomar Giltig från 2023-12-21

Genpanel PEDHEP (Pediatric Hepatology), version 21 – 169 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, 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, DYNC2LI1, EFL1, EIF2AK3, FAH, FBP1, 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, MPI, MPV17, MTTP, MYO5B, NBAS, NEK1, NGLY1, NOTCH2, NPC1, NPC2, NPHP3, NR1H4, OFD1, PGM1, PHKA2, PHKB, PHKG2, PKD1, PKD2, PKHD1, PKLR, PMM2, POLG, PPM1F, PRF1, PRKCSH, PYGL, RINT1, RPGRIP1L, SBDS, SC5D, SCYL1, SEC61B, SEC63, SEMA7A, SERPINA1, SKIV2L, SLC10A1, SLC10A2, SLC25A13, SLC25A20, SLC27A5, SLC2A2, SLC37A4, SLC51A, SLC51B, SLCO1B1, SLCO1B3, SMPD1, SRP54, TALDO1, TCTN2, TFAM, TFR2, TJP2, TMEM107, TMEM199, TMEM216, TMEM67, TRAF3IP1, TRMU, TTC21B, TTC26, TTC37, TULP3, TWNK, UGT1A1, UNC13D, UNC45A, USP53, VIPAS39, VPS33B, WDR19, WDR35, WDR83OS, XPNPEP3, ZFYVE19.