

Primer immunhiány (Primary Immunodeficiency-PID) génpanel
v:2022.11.28.

Panel tartalma 440 gén:

ACD, ACP5, ACTB, ADA, ADAM17, ADAR, AICDA, AIRE, AK2, ALPI, AP1S3, AP3B1, AP3D1, APOL1, ARHGEF1, ARPC1B, ATAD3A, ATG4A, ATM, ATP6AP1, B2M, BACH2, BCL10, BCL11B, BLM, BLNK, BRCA1, BRCA2, BRIP1, BTK, C17orf62, C19orf40, C1QA, C1QB, C1QC, C1R, C1S, C2, C2orf69, C3, C4A, C4B, C5, C6, C7, C8A, C8B, C8G, C9, CARD11, CARD14, CARD9, CASP10, CASP8, CCBE1, CD19, CD247, CD27, CD28, CD3D, CD3E, CD3G, CD40, CD40LG, CD46, CD55, CD59, CD70, CD79A, CD79B, CD81, CD8A, CDC42, CDCA7, CEBPE, CECR1, CFB, CFD, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, CFP, CFTR, CHD7, CHUK, CIB1, CIITA, CLCN7, CLPB, COPA, COPG1, CORO1A, CR2, CSF2RA, CSF2RB, CSF3R, CTC1, CTLA4, CTNNB1, CTPS1, CTSC, CXCR2, CXCR4, CYBA, CYBB, DBR1, DCLRE1C, DEF6, DIAPH1, DKC1, DNAJC21, DNASE1L3, DNASE2, DNMT3B, DOCK2, DOCK8, EFCAB4B, EFTUD1, ELANE, ELF4, EPG5, ERBB2IP, ERCC4, ERCC6L2, EXTL3, FADD, FAM105B, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FAS, FASLG, FAT4, FCGR3A, FCHO1, FCN3, FERMT1, FERMT3, FNIP1, FOXP3, FPR1, G6PC3, G6PD, GATA2, GFI1, GINS1, HAVCR2, HAX1, HCK, HELLS, HMOX1, HYOU1, ICOS, ICOSLG, IFIH1, IFNAR1, IFNAR2, IFNG, IFNGR1, IFNGR2, IGLL1, IKBKB, IKBKG, IKZF1, IKZF2, IKZF3, IL10, IL10RA, IL10RB, IL12B, IL12RB1, IL12RB2, IL17F, IL17RA, IL17RC, IL18BP, IL1RN, IL21, IL21R, IL23R, IL2RA, IL2RB, IL2RG, IL36RN, IL6R, IL6ST, IL7R, INO80, IRAK1, IRAK4, IRF2BP2, IRF3, IRF4, IRF7, IRF8, IRF9, ISG15, ITCH, ITGB2, ITK, ITPKB, JAGN1, JAK1, JAK3, KDM6A, KMT2A, KMT2D, LAMTOR2, LAT, LCK, LCP2, LIG1, LIG4, LPIN2, LRBA, LSM11, LYST, MAD2L2, MAGT1, MALT1, MAN2B2, MAP1LC3B2, MAP3K14, MAPK8, MASP2, MCM10, MCM4, MECOM, MEFV, MKL1, MLL2, MOGS, MS4A1, MSH6, MSN, MTHFD1, MVK, MYD88, MYSM1, NBAS, NBN, NCF1, NCF2, NCF4, NCKAP1L, NCSTN, NDNF2, NFAT5, NFE2L2, NFKB1, NFKB2, NFKBIA, NHEJ1, NHP2, NLRP4, NLRP1, NLRP12, NLRP3, NOD2, NOP10, NOS2, OAS1, OBFC1, OSM1, PALB2, PARN, PAX1, PDCD1, PEPD, PGM3, PIK3CD, PIK3CG, PIK3R1, PLCG2, PLEKHM1, PMS2, PNP, POLA1, POLD1, POLD2, POLE, POLE2, POLR3A, POLR3C, POLR3F, POU2AF1, PRF1, PRKCD, PRKDC, PSEN1, PSENEN, PSMB8, PSMB9, PSMG2, PSTPIP1, PTEN, PTPRC, RAB27A, RAC2, RAD51, RAD51C, RAG1, RAG2, RANBP2, RASGRP1, RBCK1, REL, RELB, RFWD3, RFX5, RFXANK, RFXAP, RHOG, RHOH, RIPK1, RLTPR, RNASEH2A, RNASEH2B, RNASEH2C, RNF168, RNF31, RORC, RPSA, RTEL1, SAMD9, SAMD9L, SAMHD1, SASH3, SBDS, SEC61A1, SEMA3E, SERPING1, SH2D1A, SH3BP2, SH3KBP1, SKIV2L, SLC29A3, SLC35C1, SLC39A7, SLC46A1, SLC7A7, SLX4, SMARCA1, SMARCA2, SNORA31, SNX10, SOCS1, SP110, SPI1, SPINK5, SPPL2A, SRP54, SRP72, STAT1, STAT2, STAT3, STAT5B, STIM1, STK4, STX11, STXBP2, SYK, TAP1, TAP2, TAPBP, TAZ, TBK1, TBX1, TBX21, TCF3, TCIRG1, TCN2, TERT, TET2, TFRC, TGFB1, TGFB2, THBD, TICAM1, TNF2, TIRAP, TLR3, TLR7, TLR8, TMC6, TMC8, TMEM173, TNFAIP3, TNFRSF11A, TNFRSF13B, TNFRSF13C, TNFRSF1A, TNFRSF4, TNFRSF9, TNFSF11, TNFSF12, TNFSF13, TOP2B, TP53, TPP2, TRAF3, TRAF3IP2, TREX1, TRIM22, TRNT1, TTC37, TTC7A, TYK2, UBE2T, UNC13D, UNC93B1, UNG, USB1, USP18, VPS13B, VPS45, WAS, WDR1, WIPF1, WRAP53, XIAP, XRCC2, ZAP70, ZBTB24, ZNF341, ZNF1.

Forrás:

Tangye SG, Al-Herz W, Bousfiha A, et al. Human Inborn Errors of Immunity: 2022 Update on the Classification from the International Union of Immunological Societies Expert Committee. *J Clin Immunol*. 2022;42(7):1473-1507. doi: 10.1007/s10875-022-01289-3