Primer immunhiány (Primary Immunodeficiency-PID) génpanel v:2025-05-28

Panel tartalma 508 gén:

ACD, ACP5, ACTB, ADA, ADA2, ADAM17, ADAR, AICDA, AIRE, AK2, ALPI, ALPK1, AP1S3, AP3B1, AP3D1, APOL1, ARF1, ARHGEF1, ARPC1B, ARPC5, ATAD3A, ATG4A, ATM, ATP6AP1, B2M, BACH2, BCL10, BCL11B, BLM, BLNK, BRCA1, BRCA2, BRIP1, BTK, C1QA, C1QB, C1QC, C1R, C1S, C2, C2orf69, C3, C4A, C4B, C5, C6, C7, C8A, C8B, C8G, C9, CARD11, CARD14, CARD9, CARMIL2, CASP10, CASP8, CBLB, CCBE1, CCR2, CD19, CD247, CD27, CD274, CD28, CD3D, CD3E, CD3G, CD40, CD40LG, CD46, CD55, CD59, CD70, CD79A, CD79B, CD81, CD8A, CDC42, CDCA7, CEBPE, CFB, CFD, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, CFP, CFTR, CHD7, CHUK, CIB1, CIITA, CLCN7, CLPB, COPA, COPG1, CORO1A, CR2, CRACR2A, CTC1, CTLA4, CTNNBL1, CTPS1, CTSC, CXCR2, CXCR4, CYBA, CYBB, CYBC1, CSF2RA, CSF2RB, CSF3R, DBF4, DBR1, DCLRE1B, DCLRE1C, DEF6, DIAPH1, DKC1, DNAJC21, DNASE1L3, DNASE2, DNMT3B, DOCK11, DOCK2, DOCK8, DPP9, DUT, EFL1, ELANE, ELF4, EPG5, ERBIN, ERCC4, ERCC6L2, ERN1, EXTL3, FAAP24, FADD, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FAS, FASLG, FAT4, FCGR3A, FCHO1, FCN3, FERMT1, FERMT3, FLT3LG, FNIP1, FOXI3, FOXN1, FOXP3, FPR1, G6PC3, G6PD, GATA2, GFI1, GIMAP6, GINS1, GINS4, GTF3A, HAVCR2, HAX1, HCK, HELLS, HMOX1, HYOU1, ICOS, ICOSLG, IFIH1, IFNAR1, IFNAR2, IFNG, IFNGR1, IFNGR2, IGHM, IGKC, IGLL1, IKBKB, IKBKE, IKBKG, IKZF1, IKZF2, IKZF3, IL10, IL10RA, IL10RB, IL12B, IL12RB2, IL17F, IL17RA, IL17RC, IL18BP, IL1R1, IL1RN, IL21, IL21R, IL23R, IL27RA, IL2RA, IL2RB, IL2RG, IL36RN, IL6R, IL6ST, IL7R, INO80, IRAK1, IRAK4, IRF1, IRF2BP2, IRF3, IRF4, IRF7, IRF8, IRF9, ISG15, ITCH, ITGB2, ITK, ITPKB, ITPR3, JAGN1, JAK1, JAK3, KARS1, KDM6A, KMT2A, KMT2D, LACC1, LAMTOR2, LAT, LCK, LCP2, LIG1, LIG4, LPIN2, LRBA, LSM11, LY96, LYN, LYST, MAD2L2, MAGT1, MALT1, MAN2B2, MAP1LC3B2, MAP3K14, MAP3K9, MAPK8, MASP2, MCM10, MCM4, MCTS1, MECOM, MEFV, MOGS, MRTFA, MS4A1, MSH6, MSN, MTHFD1, MVK, MYD88, MYSM1, NBAS, NBEAL2, NBN, NCF1, NCF2, NCF4, NCKAP1L, NCSTN, NFAT5, NFATC1, NFATC2, NFE2L2, NFKB1, NFKB2, NFKBIA, NHEJ1, NHP2, NLRC4, NLRP1, NLRP12, NLRP3, NOD2, NOP10, NOS2, NSMCE3, NUDCD3, OAS1, OAS2, ORAI1, OSTM1, OTULIN, PALB2, PARN, PAX1, PAX5, PDCD1, PEPD, PGM3, PI4KA, PIK3CD, PIK3CG, PIK3R1, PLCG1, PLCG2, PLEKHM1, PMS2, PMVK, PNP, POLA1, POLD1, POLD2, POLD3, POLE, POLE2, POLR3A, POLR3C, POLR3F, POMP, POU2AF1, PRF1, PRIM1, PRKCD, PRKDC, PSEN1, PSENEN, PSMB10, PSMB4, PSMB8, PSMB9, PSMD12, PSMG2, PSTPIP1, PTCRA, PTEN, PTPN2, PTPRC, RAB27A, RAC2, RAD50, RAD51, RAD51C, RAG1, RAG2, RANBP2, RASGRP1, RBCK1, RECQL4, REL, RELA, RELB, RFWD3, RFX5, RFXANK, RFXAP, RHBDF2, RHOG, RHOH, RIPK1, RIPK3, RMRP, RNASEH2A, RNASEH2B, RNASEH2C, RNASEL, RNF168, RNF31, RNU4ATAC, RNU7-1, RORC, RPSA, RTEL1, SAMD9, SAMD9L, SAMHD1, SASH3, SBDS, SEC61A1, SEMA3E, SERPING1, SGPL1, SH2B3, SH2D1A, SH3BP2, SH3KBP1, SHARPIN, SKIV2L, SLC19A1, SLC29A3, SLC35C1, SLC37A4, SLC39A7, SLC46A1, SLC7A7, SLX4, SMAD3, SMARCAL1, SMARCD2, SNORA31, SNX10, SOCS1, SP110, SPI1, SPINK5, SPPL2A, SRP19, SRP54, SRP72, SRPRA, STAT1,

STAT2, STAT3, STAT4, STAT5B, STAT6, STIM1, STING1, STK4, STN1, STX11, STXBP2, SYK, TAP1, TAP2, TAPBP, TBK1, TBX1, TBX21, TCF3, TCIRG1, TCN2, TERC, TERT, TET2, TFRC, TGFB1, TGFBR1, TGFBR2, THBD, TICAM1, TINF2, TIRAP, TLR3, TLR4, TLR7, TLR8, TMC6, TMC8, TNFAIP3, TNFRSF11A, TNFRSF13B, TNFRSF13C, TNFRSF1A, TNFRSF4, TNFRSF9, TNFSF11, TNFSF12, TNFSF13, TNFSF9, TOP2B, TP53, TPP2, TRAC, TRAF3, TRAF3IP2, TREX1, TRIM22, TRNT1, TTC37, TTC7A, TYK2, UBE2T, UNC13D, UNC93B1, UNG, USB1, USP18, VPS13B, VPS45, WAS, WDR1, WIPF1, WRAP53, WWTR1, XIAP, XRCC2, ZAP70, ZBTB24, ZNF341, ZNFX1

Forrás:

M. Cecilia Poli, Ivona Aksentijevich, Ahmed Aziz Bousfiha, Charlotte Cunningham-Rundles, Sophie Hambleton, Christoph Klein, Tomohiro Morio, Capucine Picard, Anne Puel, Nima Rezaei, Mikko R.J. Seppänen, Raz Somech, Helen C. Su, Kathleen E. Sullivan, Troy R. Torgerson, Isabelle Meyts, Stuart G. Tangye; Human inborn errors of immunity: 2024 update on the classification from the International Union of Immunological Societies Expert Committee. J Hum Immun 5 May 2025; 1 (1): e20250003. doi: https://doi.org/10.70962/jhi.20250003