

Seznam vyšetřovaných genů v panelu Primární imunodeficeience (PID=IEI)

V uvedených genech jsou analyzovány kódující exony a přilehlé intronové oblasti v rozsahu 20 bp

ABCB1, ACD, ACOD1, ACP5, ACTB, ADA, ADA2, ADAM17, ADAMTS3, ADAR, AICDA, AIRE, AK2, ALPI, ALPK1, ANGPT1, ANKRD26, ANKZF1, AP1M2, AP1S1, AP1S3, AP3B1, AP3D1, AP3S2, APOL1, ARHGEF1, ARPC1B, ARPC5, ARPIN, ATAD3A[†], ATG16L1, ATG4A, ATM, ATP6AP1, B2M, BACH2, BCL10, BCL11B, BLM, BLNK, BLOC1S6, BTK, C1QA, C1QB, C1QC, C1R, C1S, C2, C2orf69, C3, C5, C6, C7, C8A, C8B, C8G, C8orf34, C9, CARD11, CARD14, CARD16, CARD8, CARD9, CARMIL2, CASP10, CASP8, CASP9, CBLB, CCBE1, CCDC88A, CCR2, CD19, CD247, CD27, CD274, CD28, CD3D, CD3E, CD3G, CD4, CD40, CD40LG, CD46, CD48, CD55, CD59, CD70, CD79A, CD79B, CD81, CD8A, CDC42, CDCA7, CEBPE, CFB, CFD, CFH, CFHR1[†], CFHR2[†], CFHR3[†], CFHR4[†], CFHR5[†], CFI, CFP, CHD7, CHUK, CIB1, CIITA, CLCN7, CLPB, COG6, COL7A1, COPA, COPG1, CORO1A[†], CPN1, CR2, CRACR2A, CSF2RA, CSF2RB, CSF3R, CTC1, CTLA4, CTNBL1, CTPS1, CTSC, CWF19L2, CXCR2, CXCR4, CYBA, CYBB, CYBC1, CYCS, DAB2IP, DBF4, DBR1, DCLRE1A, DCLRE1B, DCLRE1C, DEF6, DIAPH1, DKC1, DNAJC21, DNASE1, DNASE1L3, DNASE2, DNMT3B, DOCK11, DOCK2, DOCK8, DPP9, DTNBP1, DUOX2[†], DUT, EFL1, EGFR, ELANE, ELF4, EPG5, ERBIN, ERCC6L2, ERN1, ETV6, EXTL3, F12, FAAP24, FADD, FAS, FASLG, FAT4, FCGR3A[†], FCGR3B[†], FCHO1, FCN3, FERMT1, FERMT3, FLT3LG, FNIP1, FOXI3, FOXN1, FOXP3, FPR1, FYB1, G6PC3, G6PD, GATA1, GATA2, GCC2[†], GFI1, GIMAP5, GIMAP6, GINS1, GINS4, GNAI2, GTF3A, GUCY2C, HAVCR2, HAX1, HCK, HELLS, HMOX1, HOXA3, HPS1, HPS3, HPS4, HPS5, HPS6, HS3ST6, HSPA1L, HYOU1, ICOS, ICOSLG[†], IFIH1, IFNAR1, IFNAR2, IFNG, IFNGR1, IFNGR2, IGHG2[†], IGHM, IGKC, IGLL1, IKBKB, IKBKE, IKBKG[†], IKZF1, IKZF2, IKZF3, IL10, IL10RA, IL10RB, IL12B, IL12RB1, IL12RB2, IL17A, IL17F, IL17RA, IL17RC, IL18BP, IL1R1, IL1RN, IL21, IL21R, IL22, IL23R, IL27RA, IL2RA, IL2RB, IL2RG, IL36RN, IL6, IL6R, IL6ST, IL7, IL7R, INAVA, INO80, IRAK1, IRAK4, IRF1, IRF2BP2, IRF3, IRF4, IRF5, IRF7, IRF8, IRF9, IRGM, ISG15, ITCH, ITGAM, ITGB2, ITK, ITPKB, ITPR3, JAGN1, JAK1, JAK3, KARS1, KDM6A, KMT2A, KMT2D, KNG1, LACC1, LAMTOR2, LAT, LCK, LCP2, LIG1, LIG4, LPIN2, LRBA, LSM11, LY96, LYN, LYST, MADD, MAGT1, MALT1, MAN2B2, MAP1LC3B2, MAP3K14, MAP4K1, MAPK8, MASP2, MBL2, MCM10, MCM4, MCTS1, MECOM, MEFV, MEX3B, MFAP5, MOGS, MRTFA, MS4A1, MSH6, MSN, MTHFD1, MVK, MYD88, MYH9, MYOF, MYSM1, NBAS, NBEAL2, NBN, NCF1[†], NCF2, NCF4, NCKAP1L, NCSTN, NFAT5, NFATC1, NFATC2, NFE2L2, NFIL3, NFKB1, NFKB2, NFKBIA, NFKBIB, NFKBID, NHEJ1, NHP2, NLRC4, NLRP1, NLRP12, NLRP3, NOD2, NOP10, NOS2, NOX1, NPC1, NSMCE3, NUDCD3, OAS1, OAS2, OAS3, ORAI1, OSTM1, OTULIN, PARN, PAX1, PAX5, PDCD1, PDLIM1, PEPD, PGM3, PIK3CD, PIK3CG, PIK3R1, PLA2G4A, PLCG1, PLCG2, PLEKHM1, PLG, PMS2[†], PMVK, PNP, POLA1, POLD1, POLD2, POLD3, POLE, POLE2, POLR3A, POLR3C, POLR3E, POLR3F, POMP, POU2AF1, PPM1D, PRDM1, PRF1, PRIM1, PRKCD, PRKDC, PSEN1, PSENEN, PSMA3, PSMB10, PSMB4, PSMB8, PSMB9, PSMD12, PSMG2, PSTPIP1, PTCRA, PTEN, PTPN2, PTPN22, PTPRC, RAB27A, RAC2, RAD50, RAG1, RAG2, RANBP2[†], RAP1B, RASGRP1, RBCK1, RECQL4, REL, RELA, RELB, RFX5, RFXANK, RFXAP, RHBDF2, RHOG, RHOH, RIGI, RIPK1, RIPK3, RMRP, RNASEH2A, RNASEH2B, RNASEH2C, RNASEL, RNF168, RNF186, RNF31, RNU4ATAC, RNU7-1, RORC, RPSA, RTEL1, SAMD9, SAMD9L, SAMHD1, SART3, SASH3, SBDS, SEC61A1, SEMA3E, SERPING1, SGPL1, SH2B3, SH2D1A, SH3BP2, SH3KBP1, SHARPIN, SKIC2, SKIC3, SLC19A1, SLC29A3, SLC35C1, SLC37A4, SLC39A7, SLC46A1, SLC6A20, SLC7A7, SLC9A3, SLCO2A1, SMARCAL1, SMARCD2, SMURF1, SNORA31, SNX10, SOCS1, SOCS4, SP110, SPI1, SPINK5, SPPL2A, SRC, SRP54, SRP72, SRPRA, STAT1, STAT2, STAT3, STAT4, STAT5B[†], STAT6, STEAP3, STIM1, STING1, STK4, STN1, STX11, STXBP2, STXBP3, SYK, TAFAZZIN, TAP1, TAP2, TAPBP, TBK1, TBX1, TBX2, TBX21, TCF3, TCIRG1, TCN2, TERC, TERT, TET2, TFRC, TGFB1, TGFB1R1, TGFB2R2, THBD, TICAM1, TINF2, TIRAP, TLR1, TLR10, TLR2, TLR3, TLR4, TLR5, TLR6, TLR7, TLR8, TLR9, TMC6, TMC8, TMEFF1, TNFAIP3, TNFRSF11A,

TNFRSF13B, TNFRSF13C, TNFRSF1A, TNFRSF4, TNFRSF9, TNFSF11, TNFSF12, TNFSF13, TNFSF15, TNFSF9, TOM1, TOP2B, TP53, TP63, TPP1, TPP2, TRAC, TRAF3, TRAF3IP2, TREX1, TRIM22, TRNT1, TTC7A, TYK2, UBA1, UNC119, UNC13D, UNC93B1, UNG, USB1, USP18[†], VPS13B, VPS45, WAS, WASF2, WDR1, WIPF1, WRAP53, XIAP, ZAP70, ZBTB24, ZNF341, ZNFX1

† část sekvence genů se ve vysoce homologní podobě vyskytuje jinde v genomu; sekvenční analýza těchto oblastí je obtížně hodnotitelná

Akreditovaná metoda dle: SOPA-GL-06