Prima
Cancer Genetics

MEDGENOME

Primary Immunodeficiency Gene Panel

About

Primary immunodeficiency disorder (PID) refers to a group of heterozygous disorders that result from any defects in the immune system development and poor or absent function in one or more components of the immune system. PIDs are unique from secondary immunodeficiencies that may result from other causes, such as viral or bacterial infections, malnutrition, or treatment with drugs that induce immunosuppression. Despite a heterozygous genetic component, the core features of these disorders are often very similar which makes the diagnosis and treatment complicated. Treatment option without genetic establishment may complicate further. The type and site of the specific organisms resulting the infections will help in classifying the disease further. PIDs also have non-immune manifestations.

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PID gene panel by MedGenome

- CAP accreditation for NGS panels, clinical exome, whole exome and other confirmatory testing
- Advanced sequencing technologies, automated scripts and precise bioinformatic pipelines and superior analytical performance
- Targeted sequencing represents a cost-effective approach to detect variants present in multiple/large genes in an individual
- Detects both SNPs and small InDels in the coding regions and splice site junctions
- Insilico analysis to detect CNVs (copy number variations)
- Variants are strictly classified based on ACMG guidelines
- Systemic variant analysis and clinical interpretation workflow
- Reviewed by a team of skilled scientists and clinical geneticists

Genes Covered

ACP5, ACTB, ADA, ADAM17, ADAR, AICDA, AIRE, AK2, AP1S3, AP3B1, AP3D1, APOL1, ATM, ATP6AP1, B2M, BCL10, BCL11B, BLM, BLNK, BTK, C1QA, C1QB, C1QC, C1R, C1S, C2, C3, C5, C6, C7, C8A, C8B, C8G, C9, CARD11, CARD14, CARD9, CASP10, CASP8, CCBE1, CD19, CD247, CD27, CD3D, CD3E, CD3G, CD40, CD40LG, CD46, CD55, CD59, CD79A, CD79B, CD81, CD8A, CDCA7, CEBPE, CECR1, CFB, CFD, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, CFP, CFTR, CHD7, CIITA, CLCN7, CLPB, COPA, CORO1A, CR2, CSF2RA, CSF2RB, CSF3R, CTC1, CTLA4, CTPS1, CTSC, CXCR4, CYBA, CYBB, DCLRE1C, DDX58, DKC1, DNAJC21, DNMT3B, DOCK2, DOCK8, ELANE, EPG5, ERCC6L2, EXTL3, FAAP24, FADD, FAS, FASLG, FAT4, FCGR3A, FCN3, FERMT3, FOXN1, FOXP3, FPR1, G6PC3, G6PD, GATA2, GFI1, HAX1, HELLS, HMOX1, ICOS, IFIH1, IFNAR2, IFNGR1, IFNGR2, IGHM, IGKC, IGLL1, IKBKB, IKZF1, IL10, IL10RA, IL10RB, IL12B, IL12RB1, IL17F, IL17RA, IL17RC, IL1RN, IL21R, IL2RA, IL2RG, IL36RN, IL7R, INO80, IRAK1, IRAK4, IRF2BP2, IRF3, IRF7, IRF8, ISG15, ITCH, ITGB2, ITK, JAGN1, JAK1, JAK3, KDM6A, KMT2D, LAMTOR2, LAT, LCK, LIG1, LIG4, LPIN2, LRBA, LYST, MAGT1, MALT1, MAP3K14, MASP2, MCM4, MEFV, MKL1, MOGS, MS4A1, MSH6, MSN, MTHFD1, MVK, MYD88, MYSM1, NBAS, NBN, NCF2, NCF4, NCSTN, NDNL2, NFAT5, NFKB1, NFKB2, NFKBIA, NHEJ1, NHP2, NLRC4, NLRP1, NLRP12, NLRP3, NOD2, NOP10, OBFC1, ORAI1, OSTM1, OTULIN, PARN, PEPD, PGM3, PIK3CD, PIK3R1, PLCG2, PLEKHM1, PMS2, PNP, POLA1, POLE, POLE2, PRF1, PRKCD, PRKDC, PSEN1, PSENEN, PSMB8, PSTPIP1, PTEN, PTPRC, RAB27A, RAC2, RAG1, RAG2, RANBP2, RASGRP1, RBCK1, RFX5, RFXANK, RFXAP, RHOH, RLTPR, RMRP, RNASEH2A, RNASEH2B, RNASEH2C, RNF168, RNF31, RNU4ATAC, RORC, RPSA, RTEL1, SAMD9, SAMD9L, SAMHD1, SBDS, SEMA3E, SERPING1, SH2D1A, SH3BP2, SLC29A3, SLC35C1, SLC37A4, SLC46A1, SMARCAL1, SMARCD2, SNX10, SP110, SPINK5, STAT1, STAT2, STAT3, STAT5B, STIM1, STK4, STX11, STXBP2, TAP1, TAP2, TAPBP, TAZ, TBK1, TBX1, TCF3, TCIRG1, TCN2, TERC, TERT, TFRC, THBD, TICAM1, TINF2, TIRAP, TLR3, TMC6, TMC8, TMEM173, TNFAIP3, TNFRSF11A, TNFRSF13B, TNFRSF13C, TNFRSF1A, TNFRSF4, TNFSF11, TNFSF12, TPP1, TPP2, TRAC, TRAF3, TRAF3IP2, TREX1, TRNT1, TTC37, TTC7A, TYK2, UNC13D, UNC93B1, UNG, USB1, USP18, VPS13B, VPS45, WAS, WDR1, WIPF1, WRAP53, XIAP, ZAP70, ZBTB24

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