

## IMMUNOLOJİ VE GENETİK POLİKLİNİĞİ

Kemik iliğinde meydana gelen düzensizlikler, tekrarlayan ateş, atak, otoimmün hastalık ve tedaviye yanıt vermeyen durumlar, primer bağışıklık sistemi yetmezliği, nötropeni, T ve B hücrelerinde meydana gelen çeşitli mutasyonlar sonucu fonksiyonlarında meydana gelen değişimler gibi vücudun immün sisteminin çalışma mekanizmasını değiştiren ve vücudu hastalıklara karşı savunmasız bırakan durumların analizini yapmak ve bu durum göz önüne alınarak tedavi yöntemini belirlemek için yapılan çalışmaların olduğu panellerdir.

### 1. Otoenflamatuar Sendrom Paneli Genleri;

- ✓ ACP5, ADAR, CARD14, DDX58, ELANE, IFIH1, IL1RN, IL36RN, ISG15, LPIN2, MEFV, MVK, NLRC4, NLRP1, NLRP12, NLRP3, NOD2, OTULIN, PLCG2, PRG4, PSENEN, PSMB8, PSTPIP1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, SLC29A3, TMEM173, TNFAIP3, TNFRSF1A, TREX1, TRNT1

## 2. Kemik İliği Yetmezliği Sendromu Paneli Genleri;

- ✓ ACD, ACTB, AK2, ANKRD26, AP3B1, ATM, ATR, BLM, BLOC1S3, BLOC1S6, BRAF, BRCA1, BRCA2, BRIP1, CBL, CDKN2A, CEBPA, CLPB, CSF2RA, CSF3R, CTC1, CTSC, CXCR4, DDX41, DKC1, DNAJC21, DNASE2, DTNBP1, EFL1, ELANE, EPCAM, ERCC4, ERCC6L2, ETV6, FADD, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FAS, FASLG, G6PC3, GATA1, GATA2, GFI1, GINS1, HAX1, HPS1, HPS3, HPS4, HPS5, HPS6, HRAS, IFNGR2, IKZF1, ITK, JAGN1, KRAS, LAMTOR2, LYST, MAGT1, MAP2K1, MAP2K2, MKL1, MLH1, MPL, MSH2, MSH6, MYO5A, NBN, NF1, NHP2, NOP10, NRAS, PALB2, PAX5, PGM3, PMS2, PRF1, PTPN11, RAB27A, RAC2, RAD51C, RBM8A, RECQL4, RIT1, RPL11, RPL15, RPL27, RPL31, RPL35A, RPL5, RPS10, RPS19, RPS24, RPS26, RPS28, RPS29, RPS7, RTEL1, RUNX1, SAMD9, SAMD9L, SBDS, SH2D1A, SLC37A4, SLX4, SMARCD2, SOS1, SRP54, SRP72, STX11, STXBP2, TERC, TERT, THPO, TINF2, TP53, UBE2T, UNC13D, USB1, VPS13B, VPS45, WAS, WDR1, WIPF1, WRAP53, XIAP, XRCC2

## 3. Kronik Granüloamatöz Hastalık Paneli Genleri;

- ✓ C17ORF62, CYBA, CYBB, G6PD, NCF1, NCF2, NCF4, NOD2

**4. Komplementer Sistem Bozukluğu Paneli Genleri;**

- ✓ ADIPOQ, ADIPOR1, ADIPOR2, ARMC4, C11ORF70, C1QA, C1QB, C1QBP, C1QC, C1S, C2, C3, C3AR1, C4BPA, C4BPB, C5, C5AR1, C5AR2, C6, C7, C8A, C8B, C8G, C9, CCDC103, CCDC114, CCDC39, CCDC40, CCDC65, CCNO, CD46, CD55, CD59, CD93, CFB, CFD, CFH, CFI, CFP, CLU, COLEC11, CR2, CRP, DGKE, DAAAF1, DAAAF2, DAAAF3, DAAAF5, DNAH11, DNAH5, DNAH9, DNAI1, DNAI2, DNAL1, DRC1, DYX1C1, FCN1, FCN2, FCN3, GAS2L2, HYDIN, LRRC6, MASP1, MASP2, MAT2A, MCIDAS, NME8, OFD1, PIGA, PTX3, RSPH1, RSPH4A, RSPH9, SERPING1, SPAG1, STK36, THBD, VSIG4, VTN, ZMYND10

**5. Konjenital Nötropeni Paneli Genleri;**

- ✓ ACTB, CLPB, CSF2RA, CSF3R, CTSC, EFL1, ELANE, G6PC3, GATA2, GF11, GINS1, HAX1, IFNGR2, JAGN1, LAMTOR2, LYST, MKL1, PGM3, RAC2, SBDS, SLC37A4, SMARCD2, SRP54, SRP72, VPS13B, VPS45, WAS, WDR1

**6. Diskeratozis Konjenita Paneli Genleri;**

- ✓ AK2, CTC1, DKC1, NHP2, NOP10, PARN, RTEL1, TERC, TERT, TINF2, USB1, WRAP53

**7. Hemafagositik Lenfositik Paneli Genleri;**

- ✓ FADD, FAS, FASLG, ITK, LYST, MAGT1, MYO5A, PRF1, RAB27A, RECQL4, SH2D1A, STX11, STXBP2, UNC13D, XIAP

**8. Primer İmmun  
Yetmezlik (PID)  
ve Primer Siliyer  
Diskinezi (PCD)  
Paneli Genleri;**

✓ ACD, ACP5, ACTB, ADA, ADAM17, ADAR, AICDA, AIRE, AK2, ALPI, AP3B1, ARHGEF1, ARMC4, ARPC1B, ATM, ATP6AP1, BACH2, BCL10, BCL11B, BLM, BLNK, BTK, C11ORF70, C17ORF62, C1QA, C1QB, C1QC, C1S, C2, C21ORF59, C3, CARD11, CARD14, CARD9, CASP10, CASP8, CCDC103, CCDC114, CCDC39, CCDC40, CCDC65, CCNO, CD19, CD247, CD27, CD3D, CD3E, CD3G, CD40, CD40LG, CD46, CD55, CD59, CD70, CD79A, CD79B, CD81, CD8A, CDCA7, CDK9, CEBPE, CECR1, CENPF, CFB, CFD, CFH, CFI, CFP, CFTR, CHD7, CIITA, CLCN7, CLPB, COLEC11, COPA, CORO1A, CR2, CSF2RA, CSF2RB, CSF3R, CTC1, CTLA4, CTPS1, CTSC, CXCR4, CYBA, CYBB, DBR1, DCLRE1C, DDX58, DGKE, DKC1, DNAAF1, DNAAF2, DNAAF3, DNAAF5, DNAH1, DNAH11, DNAH5, DNAH9, DNAI1, DNAI2, DNAJC21, DNAL1, DNASE2, DNMT3B, DOCK2, DOCK8, DRC1, DYX1C1, EFL1, ELANE, EPG5, ERCC6L2, EXTL3, FADD, FAS, FASLG, FCHO1, FERMT3, FOXN1, FOXP3, G6PC3, G6PD, GAS2L2, GAS8, GATA2, GFII, GINS1, HAX1, HELLS, HYDIN, HYOU1, ICOS, IFIH1, IFNAR2, IFNGR1, IFNGR2, IGLL1, IKBKB, IKZF1, IL10, IL10RA, IL10RB, IL12B, IL12RB1, IL17RA, IL17RC, IL1RN, IL21, IL21R, IL23R, IL2RA, IL2RB, IL2RG, IL36RN, IL6ST, IL7R, INVS, IRAK4, IRF2BP2, IRF8, ISG15, ITGB2, ITK, JAGN1, JAK1, JAK3, KRAS, LAMTOR2, LAT, LCK, LIG1, LIG4, LPIN2, LRBA, LRRC6, LYST, MAGT1, MALT1, MAP3K14, MASP1, MCIDAS, MEFV, MKL1, MOGS, MRE11A, MSN, MTHFD1, MVK, MYD88, MYO5A, NBN, NCF1, NCF2, NCF4, NCSTN, NFKB1, NFKB2, NFKBIA, NHEJ1, NHP2, NLRC4, NLRP1, NLRP12, NLRP3, NME8, NOD2, NOP10, NRAS, NSMCE3, OFD1, ORAI1, OTULIN, PARN, PEPD, PGM3, PIGA, PIH1D3, PIK3CD, PIK3R1, PLCG2, PMS2, PNP, POLD1, POLE, POLE2, POMP, PRF1, PRG4, PRKCD, PRKDC, PSENEN, PSMB8, PSTPIP1, PTPRC, RAB27A, RAC2, RAG1, RAG2, RASGRP1, RBCK1, RECQL4, RELA, RFX5, RFXANK, RFXAP, RHOH, RIPK1, RLTPR, RMRP, RNASEH2A, RNASEH2B, RNASEH2C, RNF168, RNF31, RNU4ATAC, RORC, RPGR, RPSA, RSPH1, RSPH3, RSPH4A, RSPH9, RTEL1, SAMD9, SAMD9L, SAMHD1, SBDS, SERPING1, SH2D1A, SLC29A3, SLC35C1, SLC37A4, SLC39A7, SLC46A1, SLC7A7, SMARCAL1, SMARCD2, SP110, SPAG1, SPINK5, SPPL2A, SRP54, SRP72, STAT1, STAT2, STAT3, STAT5B, STIM1, STK36, STK4, STX11, STXBP2, TAP1, TAP2, TAPBP, TBX1, TCF3, TCN2, TERC, TERT, TFRC, TGFB1, THBD, TINF2, TMC6, TMC8, TMEM173, TNFAIP3, TNFRSF13B, TNFRSF1A, TNFRSF4, TNFRSF9, TRAF3IP2, TREX1, TRNT1, TTC37, TTC7A, TYK2, UNC119, UNC13D, UNC93B1, UNG, USB1, USP18, VPS13B, VPS45, WAS, WDR1, WIPF1, WRAP53, XIAP, ZAP70, ZBTB24, ZMYND10, ZNF341

## 9. Primer İmmün Yetmezlik Paneli Genleri;

- ✓ ACD, ACP5, ACTB, ADA, ADAM17, ADAR, AICDA, AIRE, AK2, ALPI, AP3B1, ARHGEF1, ARPC1B, ATM, ATP6AP1, BACH2, BCL10, BCL11B, BLM, BLNK, BTK, C17ORF62, C1QA, C1QB, C1QC, C1S, C2, C3, CARD11, CARD14, CARD9, CASP10, CASP8, CD19, CD247, CD27, CD3D, CD3E, CD3G, CD40, CD40LG, CD46, CD55, CD59, CD70, CD79A, CD79B, CD81, CD8A, CDCA7, CDK9, CEBPE, CECR1, CFB, CFD, CFH, CFI, CFP, CFTR, CHD7, CIITA, CLCN7, CLPB, COLEC11, COPA, CORO1A, CR2, CSF2RA, CSF2RB, CSF3R, CTC1, CTLA4, CTPS1, CTSC, CXCR4, CYBA, CYBB, DBR1, DCLRE1C, DDX58, DGKE, DKC1, DNAJC21, DNASE2, DNMT3B, DOCK2, DOCK8, EFL1, ELANE, EPG5, ERCC6L2, EXTL3, FADD, FAS, FASLG, FCHO1, FERMT3, FOXN1, FOXP3, G6PC3, G6PD, GATA2, GFI1, GINS1, HAX1, HELLS, HYOU1, ICOS, IFIH1, IFNAR2, IFNGR1, IFNGR2, IGLL1, IKBKB, IKZF1, IL10, IL10RA, IL10RB, IL12B, IL12RB1, IL17RA, IL17RC, IL1RN, IL21, IL21R, IL23R, IL2RA, IL2RB, IL2RG, IL36RN, IL6ST, IL7R, IRAK4, IRF2BP2, IRF8, ISG15, ITGB2, ITK, JAGN1, JAK1, JAK3, KRAS, LAMTOR2, LAT, LCK, LIG1, LIG4, LPIN2, LRBA, LYST, MAGT1, MALT1, MAP3K14, MASP1, MEFV, MKL1, MOGS, MRE11A, MSN, MTHFD1, MVK, MYD88, MYO5A, NBN, NCF1, NCF2, NCF4, NCSTN, NFKB1, NFKB2, NFKBIA, NHEJ1, NHP2, NLRC4, NLRP1, NLRP12, NLRP3, NOD2, NOP10, NRAS, NSMCE3, OFD1, ORAI1, OTULIN, PARN, PEPD, PGM3, PIGA, PIK3CD, PIK3R1, PLCG2, PMS2, PNP, POLD1, POLE, POLE2, POMP, PRF1, PRG4, PRKCD, PRKDC, PSENEN, PSMB8, PSTPIP1, PTPRC, RAB27A, RAC2, RAG1, RAG2, RASGRP1, RBCK1, RECQL4, RELA, RFX5, RFXANK, RFXAP, RHOH, RIPK1, RLTPR, RMRP, RNASEH2A, RNASEH2B, RNASEH2C, RNF168, RNF31, RNU4ATAC, RORC, RPSA, RTEL1, SAMD9, SAMD9L, SAMHD1, SBDS, SERPING1, SH2D1A, SLC29A3, SLC35C1, SLC37A4, SLC39A7, SLC46A1, SLC7A7, SMARCAL1, SMARCD2, SP110, SPINK5, SPPL2A, SRP54, SRP72, STAT1, STAT2, STAT3, STAT5B, STIM1, STK4, STX11, STXBP2, TAP1, TAP2, TAPBP, TBX1, TCF3, TCN2, TERC, TERT, TFRC, TGFB1, THBD, TINF2, TMC6, TMC8, TMEM173, TNFAIP3, TNFRSF13B, TNFRSF1A, TNFRSF4, TNFRSF9, TRAF3IP2, TREX1, TRNT1, TTC37, TTC7A, TYK2, UNC119, UNC13D, UNC93B1, UNG, USB1, USP18, VPS13B, VPS45, WAS, WDR1, WIPF1, WRAP53, XIAP, ZAP70, ZBTB24, ZNF341

□ **10. Ağır Kombine  
İmmün Yetmezlik  
Paneli Genleri;**

- ✓ ADA, AK2, ATM, BCL11B, BLM, CARD11, CD247, CD27, CD3D, CD3E, CD3G, CD40, CD40LG, CD8A, CIITA, CORO1A, DCLRE1C, DNMT3B, DOCK8, EPG5, FOXP1, IFNGR1, IKBKB, IL12RB1, IL2RA, IL2RG, IL7R, IRF8, ITGB2, ITK, JAK3, LAT, LCK, LIG4, LRBA, MAGT1, MALT1, MAP3K14, MSN, NHEJ1, NSMCE3, ORAI1, PARN, PGM3, PIK3CD, PMS2, PNP, POLE, POLE2, PRKDC, PTPRC, RAC2, RAG1, RAG2, RFX5, RFXANK, RFXAP, RHOH, RMRP, RTEL1, SH2D1A, SMARCA1, SP110, SPINK5, STAT1, STAT2, STAT3, STAT5B, STIM1, STK4, TAP1, TAP2, TAPBP, TBX1, TFRC, TNFRSF4, TYK2, UNC119, WAS, ZAP70