

## HEMATOLOJİ VE GENETİK POLİKLİNİĞİ

Kansızlık, kemik iliğindeki hücrelerde meydana gelen değişimler, eritroplastopeni ile karakterize aplastik anemi, trombosit aregasyonu durumu, eritrositlerin protein kaybına bağlı lipid yapısının ve yüzey alanının bozulması gibi hematolojiyi ilgilendiren çoğu acil müdahale gerektiren durumları önceden raporlamak ve tedaviyi kolaylaştırmak için yapılan çalışmaların olduğu panellerdir.

### 1. Anemi Paneli Genleri;

- ✓ ABCB7, ADAMTS13, AK1, ALAS2, AMN, ANK1, ATM, ATR, ATRX, BLM, BRCA2, BRIP1, C15ORF41, CDAN1, CLCN7, CUBN , CYB5R3, DHF, DNAJC21, DNASE2, EFL1 , EPB42, ERCC4, FANCA, FANCB, FANCC, FANCD2 , FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, G6PD, GATA1, GCLC, GPI, GSS, HBA1 , HBA2 , HBB, HFE, KIF23, KLF1, LPIN2, MTR, NBN, NT5C3A, PALB2, PC, PDHA1, PDHX, PIEZO1, PKLR, PUS1, RAD51C, REN, RHAG, RPL11, RPL15 , RPL27, RPL31, RPL35A, RPL5, RPS10, RPS19, RPS24, RPS26, RPS28, RPS29, RPS7, SBDS , SEC23B, SLC11A2, SLC19A2, SLC25A38, SLC4A1, SLX4, SPTA1, SPTB, SRP54, TCN2, TF, THBD, TMPRSS6, TPI1, XRCC2, YARS2

### 2. Kanama Bozukluğu / Koagülopati Paneli Genleri;

- ✓ ABCG5, ABCG8, ACTN1, ADAMTS13, ANKRD26, AP3B1, ARPC1B, BLOC1S3, BLOC1S6, CYCS , DTNBP1, EFL1 , ETV6, F10, F11, F12, F13A1, F2, F5, F7, F8 , F9, FGA, FGB, FGG, FLI1, FLNA, FYB, GATA1, GBA , GFII1B, GGCX, GP1BA, GP1BB, GP9, HOXA11, HPS, HPS3, HPS4, HPS5, HPS6, ITGA2, ITGA2B, ITGB3, LMAN1, MASTL, MCFD2, MECOM, MPL, MYH9, NBEAL2, P2RY12, PRKACG, PROC, PROS1 , RASGRP2, RBM8A , RUNX1, SERPINC1, SERPINF2, SLFN14, SRC, SRP54, TBXA2R, THBD, THPO, TUBB1, VKORC1, VWF, WAS, WIPF1

### 3. Kemik İliği Yetmezliği Sendromu Paneli Genleri;

- ✓ ACD, ACTB, AK2, ANKRD26, AP3B1, ATM, ATR, BL, 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, GFII, 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

### 4. Pıhtılaşma Faktörü Eksikliği Paneli Genleri;

- ✓ F10, F11, F12, F13A1, F2, F5, F7, F8, F9, FGA, FGB, FGG, GGCX, LMAN1, VKORC1, VWF

## 5. Kapsamlı Hematoloji ve Kalıtsal Kanser Paneli Genleri;

- ✓ ABCA3, ABCB7, ABCG5, ABCG8, ACD, ACTB, ACTN1, ADAMTS13, AIP, AK1, AK2, ALAS2, ALK, AMN, ANK1, ANKRD26, AP3B1, AP3D1, APC, ARPC1B, ATM, ATR, ATRX, AXIN2, BAP1, BARD1, BLM, BLOC1S3, BLOC1S6, BMPR1A, BRAF, BRCA1, BRCA2, BRIP1, BUB1B, C15ORF41, CBL, CD70, CDAN1, CDC42, CDC73, CDH1, CDK4, CDKN1B, CDKN1C, CDKN2A, CEBPA, CEP57, CHEK2, CLCN7, CLPB, CSF2RA, CSF3R, CTC1, CTSC, CUBN, CXCR4, CYB5R3, CYCS, CYLD, DDB2, DDX41, DHFR, DICER1, DIS3L2, DKC1, DNAJC21, DNASE2, DTNBP1, EFL1, EGFR, EGLN1, ELANE, EPAS1, EPB41, EPB42, EPCAM, EPOR, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6L2, ETV6, EXO1, EXT1, EXT2, EZH2, F10, F11, F12, F13A1, F13B, F2, F5, F7, F8, F9, FADD, FAM111B, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FAS, FASLG, FGA, FGB, FGG, FH, FLCN, FLI1, FLNA, FYB, G6PC3, G6PD, GALNT12, GATA1, GATA2, GBA, GCLC, GFII1, GFII2, GGCX, GINS1, GP1BA, GP1BB, GP9, GPC3, GPI, GPR143, GREM1, GSS, HAX1, HBA1, HBA2, HBB, HFE, HNF1A, HOXA11, HOXB13, HPS1, HPS3, HPS4, HPS5, HPS6, HRAS, IFNGR2, IKZF1, ITGA2, ITGA2B, ITGB3, ITK, JAGN1, JAK2, KIF23, KIT, KITLG, KLF1, KRAS, LAMTOR2, LMAN1, LPIN2, LYST, LZTR1, MAGT1, MAP2K1, MAP2K2, MASTL, MAX, MCFD2, MECOM, MEN1, MET, MIF, MKL1, MLH1, MLH3, MPL, MRE11A, MSH2, MSH3, MSH6, MTHFD1, MTR, MUTYH, MYH9, MYO5A, NBEAL2, NBN, NF1, NF2, NHP2, NOP10, NRAS, NSD1, NSUN2, NT5C3A, NTHL1, OCA2, P2RY12, PALB2, PARN, PAX5, PC, PDGFRA, PDHA1, PDHX, PGM3, PHOX2B, PIEZO1, PKLR, PMS1, PMS2, POLD1, POLE, POLH, POT1, PPM1D, PRF1, PRKACG, PRKAR1A, PROC, PROS1, PTCH1, PTEN, PTPN11, PUS1, RAB27A, RAC2, RAD50, RAD51C, RAD51D, RAF1, RASA2, RASGRP2, RB1, RBM8A, RECQL, RECQL4, REN, REST, RET, RHAG, RHBDF2, RIT1, RNF168, RPL11, RPL15, RPL27, RPL31, RPL35A, RPL5, RPS10, RPS19, RPS20, RPS24, RPS26, RPS28, RPS29, RPS7, RRAS, RTEL1, RUNX1, SAMD9, SAMD9L, SBDS, SDHA, SDHAF2, SDHB, SDHC, SDHD, SEC23B, SERPINC1, SERPINF2, SFTPB, SFTPC, SH2D1A, SHOC2, SLC11A2, SLC19A2, SLC25A38, SLC37A4, SLC45A, SLC46A1, SLC4A1, SLFN14, SLX4, SMAD4, SMARCA4, SMARCB1, SMARCD2, SOS1, SOS2, SPRED1, SPTA1, SPTB, SRC, SRP54, SRP72, STK11, STX11, STXBP2, SUFU, TBXA2R, TCN2, TERC, TERT, TF, THBD, THPO, TINF2, TMEM127, TMPRSS6, TP53, TPII, TRNT1, TSC1, TSC2, TUBB1, TYR, TYRP1, UBE2T, UNC13D, USB1, VHL, VKORC1, VPS13B, VPS45, VWF, WAS, WDR1, WIPF1, WRAP53, WRN, WT1, XIAP, XPA, XPC, XRCC2, YARS2

## 6. Konjenital Nötropeni Paneli Genleri;

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

## 7. Kapsamlı Hematoloji Panelleri Genleri;

- ✓ ABCA3, ABCB7, ABCG5, ABCG8, ACD, ACTB, ACTN1, ADAMTS13, AK1, AK2, ALAS2, AMN, ANK1, ANKRD26, AP3B1, AP3D1, ARPC1B, ATM, ATR, ATRX, BLM, BLOC1S3, BLOC1S6, BRAF, BRCA1, BRCA2, BRIP1, C15ORF41, CBL, CDAN1, CDC42, CDKN2A, CEBPA, CLCN7, CLPB, CSF2RA, CSF3R, CTC1, CTSC, CUBN, CXCR4, CYB5R3, CYCS, DDX41, DHFR, DKC1, DNAJC21, DNASE2, DTNBP1, EFL1, EGLN1, ELANE, EPAS1, EPB41, EPB42, EPCAM, EPOR, ERCC4, ERCC6L2, ETV6, F10, F1, F12, F13A1, F13B, F2, F5, F7, F8, F9, FADD, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FAS, FASLG, FGA, FGB, FGG, FLI1, FLNA, FYB, G6PC3, G6PD, GATA1, GATA2, GBA, GCLC, GFII, GFIIIB, GGCX, GINS1, GP1BA, GP1BB, GP9, GPI, GPR143, GSS, HAX1, HBA1, HBA2, HBB, HFE, HOXA11, HPS1, HPS3, HPS4, HPS5, HPS6, HRAS, IFNGR2, IKZF1, ITGA2, ITGA2B, ITGB3, ITK, JAGN1, JAK2, KIF23, KLF1, KRAS, LAMTOR2, LMAN1, LPIN2, LYST, MAGT1, MAP2K1, MAP2K2, MASTL, MCFD2, MECOM, MKL1, MLH1, MPL, MSH2, MSH6, MTHFD1, MTR, MYH9, MYO5A, NBEAL2, NBN, NF1, NHP2, NOP10, NRAS, NT5C3A, OCA2, P2RY12, PALB2, PARN, PAX5, PC, PDHA1, PDHX, PGM3, PIEZO1, PKLR, PMS2, PRF1, PRKACG, PROC, PROS1, PTPN11, PUS1, RAB27A, RAC2, RAD51C, RASGRP2, RBM8A, RECQL4, REN, RHAG, RIT1, RNF168, RPL11, RPL15, RPL27, RPL31, RPL35A, RPL5, RPS10, RPS19, RPS24, RPS26, RPS28, RPS29, RPS7, RTEL1, RUNX1, SAMD9, SAMD9L, SBDS, SEC23B, SERPINC1, SERPINF2, SFTPB, SFTPC, SH2D1A, SLC11A2, SLC19A2, SLC25A38, SLC37A4, SLC45A2, SLC46A1, SLC4A1, SLFN14, SLX4, SMARCD2, SOS1, SPTA1, SPTB, SRC, SRP54, SRP72, STX11, STXBP2, TBXA2R, TCN2, TERC, TERT, TF, THBD, THPO, TINF2, TMPRSS6, TP53, TPII, TRNT1, TUBB1, TYR, TYRP1, UBE2T, UNC13D, USB1, VKORC1, VPS13B, VPS45, VWF, WAS, WDR1, WIPF1, WRAP53, XIAP, XRCC2, YARS2



**8. Diamond-Blackfan Anemisi Paneli Genleri;**

- ✓ GATA1, RPL11, RPL15, RPL27, RPL31, RPL35A, RPL5, RPS10, RPS19, RPS24, RPS26, RPS28, RPS29, RPS7



**9. Diskeratozis Konjenita Paneli Genleri;**

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



**10. Fanconi Anemisi Paneli Genleri;**

- ✓ ATM, ATR, BLM, BRCA2, BRIP1, CXCR4, ERCC4, ERCC6L2, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, NBN, PALB2, RAD51C, SLX4, UBE2T, XRCC2



**11. Hemafagositik Lenfositosis Paneli Genleri;**

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



**12. Kalıtsal Lösemi Paneli Genleri;**

- ✓ ANKRD26, ATM, BLM, BRAF, BRCA1, BRCA2, CBL, CDKN2A, CEBPA, DDX41, DKC1, EFL1, EPCAM, ETV6, FANCA, GATA2, HRAS, IKZF1, KRAS, MAP2K1, MAP2K2, MLH1, MSH2, MSH6, NBN, NF1, NRAS, PAX5, PMS2, PTPN11, RIT1, RUNX1, SAMD9, SAMD9L, SBDS, SOS1, SRP72, TERC, TERT, TINF2, TP53



**13. Hermansky-Pudlak  
Sendromu Paneli  
Genleri;**

✓ ABCA3, AP3B1, AP3D1, BLOC1S3, BLOC1S6, DKC1, DTNBP1, GPR143, HPS1 , HPS3, HPS4, HPS5, HPS6, LYST, OCA2, SFTPB, SFTPC, SLC45A2, TERC, TERT, TINF2, TYR, TYRP1



**14. Trombosit  
Fonksiyon Bozukluđu  
Paneli Genleri;**

✓ AP3B1, ARPC1B, BLOC1S3, BLOC1S6, DTNBP1, GBA, GP1BA, GP1BB, GP9, HPS1, HPS3, HPS4, HPS5, HPS6, ITGA2B, ITGB3, NBEAL2, P2RY12, RASGRP2, RUNX1, TBXA2R, THPO, WIPF1



**15. Eritrosit Membran  
Bozukluđu Paneli  
Genleri;**

✓ ANK1, EPB41, EPB42, RHAG, SLC4A1, SPTA1, SPTB



**16. Trombositopeni  
Paneli Genleri;**

✓ ABCG5, ABCG8, ACTN1, ADAMTS13, ANKRD26, ARPC1B, CYCS, EFL1, ETV6, FLI1, FLNA, FYB, GATA1, GBA, GF11B, GP1BA, GP1BB, GP9, HOXA11, ITGA2, ITGA2B, ITGB3, MASTL, MECOM, MPL, MYH9, NBEAL2, PRKACG, RBM8A, RUNX1, SLFN14, SRC, SRP54, THBD, TUBB1, WAS, WIPF1