KARDİYOLOJİ POLİKLİNİĞİ

Doğuştan meydana gelen kalp anomalileri, belirli bölgelerde yer alan aort hastalıkları, kalp kasında meydana gelen değişimler, kan damarını etkileyen kalıtsal bağ dokusu bozuklukları, aşırı genişlemiş kapiller ve venüllere bağlı kanamalar gibi kardiyolojik durumlara cevap bulmak ve tedaviyi kolaylaştırıp yenidoğan ünitelerinde erken teşhis oluşturabilmek için yapılan çalışmaların olduğu panellerdir.

1. Kalıtsal Torasik Aorta Hastalıkları Paneli Genleri;

- ✓ ABCC6, ABL1, ACTA2, ADAMTS10, ADAMTS17, ADAMTS2, ADAMTSL4, ALDH18A1, ATP7A, B3GAT3, BGN, CBS, COL1A1, COL1A2, COL2A1, COL3A1, COL4A5, COL5A1, COL5A2, COLGALT1, EFEMP2, ELN, ENPP1, FBLN5, FBN1, FBN2, FKBP14, FLNA, FOXE3, GATA5, HCN4, LOX, MAT2A, MED12, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PRKG1, SKI, SLC2A10, SLC39A13, SMAD2, SMAD3, SMAD4, SMAD6, TGFB2, TGFB3, TGFBR1, TGFBR2, ZDHHC9

2. Aritmi (non-LQTS) Paneli Genleri;

- ✓ ABCC9, AKAP9, ANK2, BAG3, CACNA1C, CACNB2, CALM1, CALM2, CALM3, CASQ2, CAV3, CDH2, CTNNA3, DBH, DES, DSC2, DSG2, DSP, FLNC, GATA6, HADHA, HCN4, JUP, KCNA5, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ5, KCNQ1, LDB3, LEMD2, LMNA, MYH6, MYH7, MYL4, NKX2-5, NOS1AP, NUP155, PKP2, PLN, PPA2, RYR2, SALL4, SCN10A, SCN1B, SCN3B, SCN5A, TBX5, TECRL, TGFB3, TMEM43, TNNI3, TNNI3K, TNNT2, TRDN, TRPM4, TTN

3. Aritmojenik Sağ Ventriküler Kardiyomiyopati (ARVC) Paneli Genleri;

- ✓ BAG3, CDH2, CTNNA3, DES, DSC2, DSG2, DSP, FLNC, JUP, LDB3, LEMD2, LMNA, MYH7, NKX2-5, PKP2, PLN, RYR2, SCN5A, TGFB3, TMEM43, TTN

4. Atriyal Fibrilasyon Paneli Genleri;

✓ CACNB2, GATA6, HCN4, KCNA5, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ5, KCNQ1, LDB3, LMNA, NUP155, RYR2, SCN10A, SCN1B, SCN3B, SCN5A, TBX5

5. Brugada Sendromu Paneli Genleri;

✓ CACNA1C, CACNB2, HCN4, KCNH2, SCN3B, SCN5A, TRPM4

6. Kardiyomiyopati Paneli Genleri;

✓ AARS2, ABCC6, ABCC9, ACAD9, ACADVL, ACTA1, ACTC1, ACTN2, AGK, AGL, ALMS1, ALPK3, ANO5, APOA1, BAG3, BRAF, CALR3, CAPN3, CASQ2 , CASZ1 , CBL , CDH2 , CHRM2 , COX15 , CPT2 , CRYAB , CSRP3, CTNNA3, DBH, DES, DMD, DNAJC19, DOLK, DPM3, DSC2, DSG2, DSP, DTNA, DYSF, EEF1A2, ELAC2, EMD, EPG5, ETFA, ETFB, ETFDH, FBXL4, FBXO32, FHL1, FHOD3, FKRP, FKTN, FLNC, FOXD4, FOXRED1, FXN , GAA, GATA4 , GATA6, GATAD1, GATC , GBE1, GFM1, GLA, GLB1, GMPPB, GSK3B, GTPBP3, GUSB , HADHA, HAND1, HCN4, HFE, HRAS, IDUA, ILK, ISPD, JPH2, JUP, KLHL24, KRAS , LAMA2, LAMP2, LARGE, LDB3, LEMD2, LMNA, LMOD2, LRRC10, LZTR1, MAP2K1, MAP2K2, MAP3K8, MIPEP, MLYCD, MT-ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MT-RNR1, MT-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY, MTO1 , MYBPC3, MYBPHL, MYH6, MYH7, MYL2, MYL3, MYL4, MYOT, MYPN, MYRF, NDUFAF2, NEXN, NF1 , NKX2-5, NONO, NRAP, NRAS, PCCA, PCCB , PKP2 , PLEC, PLEKHM2, PLN, PNPLA2, PPA2, PPCS, PPP1CB, PRDM16, PRKAG2, PTPN11, QRSL1, RAF1, RASA2, RBCK1, RBM20, RIT1, RMND1, RRAS, RYR2, SCN5A, SCNN1B, SCNN1G, SCO1, SCO2, SDHA , SELENON , SGCA, SGCB, SGCD, SGCG, SHOC2, SLC22A5, SLC25A20, SLC25A4, SMCHD1, SOS1, SOS2, SPEG, SPRED1, TAB2, TAZ, TBX20 , TBX5, TCAP, TGFB3, TMEM43, TMEM70, TNNC1, TNNI3, TNNI3K, TNNT2, TOR1AIP1, TPM1, TRIM32, TSFM, TTN , TTR, VCL, VCP, VPS13A, XK

7. Katekolaminerjik Polimorfik Ventriküler Taşikardi (CPVT) Paneli Genleri;

- ✓ ANK2, CALM1 , CALM2, CALM3, CASQ2, KCNJ2, LMNA, RYR2, TECRL, TRDN

8. Konjenital Yapısal Kalp Hastalığı Paneli Genleri;

- ✓ ABL1, ACTA2, ACTB, ACTC1, ACTG1, ACVR1, ACVR2B, ADAMTS10, ADAMTS17, AFF4, AMMECR1, ARHGAP31, ARID1A, ARID1B, B3GAT3 , BCOR, BMPR2, BRAF , C12ORF57, CBL, CDK13, CDK9, CFAP53, CHD4, CHD7, CHRM2, CREBBP, CRELD1, CTC1, DHCR7, DLL4, DOCK6, EFTUD2, EHMT1, EIF2AK4, ELN, ENG, EOGT, EP300, EVC, EVC2, FLNA, FOXC1, FOXF1, FOXH1, FOXP1, GATA4 , GATA5, GATA6, GDF1, GJA1 , GJA5, GPC3, HAND1, HAND2, HDAC8, HNRNPK, HOXA1, HRAS, JAG1, KDM6A, KMT2D, KRAS , KYNU, LEFTY2 , MED12, MED13L, MEIS2, MMP21, MYCN, MYO18B, MYRF, NAA15, NF1 , NIPBL, NKX2-5, NKX2-6, NODAL, NONO, NOTCH1, NOTCH2 , NR2F2, NSD1, PITX2, PKD1L1, PPP1CB, PRDM6, PRKD1, PTPN11, PUF60, RAB23, RAF1, RBM10, RERE , RIT1, SALL4, SMARCB1, SMC1A, SMC3, SOS1, SOS2, STAG2, STRA6, TAB2, TBX1, TBX20, TBX5, TFAP2B, TGDS, TLL1, TMEM94, ZEB2 , ZFPM2, ZIC3

9. Kapsamlı Kardiyoloji Paneli Genleri;

✓ AARS2, ABCC6, ABCC9, ACAD9, ACADVL, ACTA1, ACTA2, ACTC1, ACTN2, AGK , AGL, AGPAT2, AKAP9, ALMS1 , ALPK3, ANK2, ANO5, APOA1, ATPAF2, BAG3, BRAF, CACNA1C , CACNB2, CALM1 , CALM2, CALM3, CALR3, CAPN3, CASQ2, CASZ1, CAV3, CBL, CDH2, CHRM2, COX15, CPT2, CRYAB, CSRP3, CTNNA3, DBH, DES, DMD, DNAJC19, DOLK, DPM3, DSC2, DSG2, DSP, DTNA, DYSF, EEF1A2, ELAC2, EMD, ENPP1, EPG5, ETFA, ETFB, ETFDH, FAH, FBXL4, FBXO32, FHL1 , FHOD3, FKRP, FKTN, FLNC , FOXD4 , FOXRED1, FXN , GAA, GATA4 , GATA5, GATA6, GATAD1, GATC , GBE1, GFM1, GLA, GLB1, GMPPB, GSK3B, GTPBP3, GUSB , HADHA, HAND1, HAND2, HCN4, HFE, HRAS, IDUA, ILK, ISPD, JPH2, JUP, KCNA5, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ5, KCNQ1, KLHL24, KRAS , LAMA2, LAMP2, LARGE, LDB3, LEMD2, LMNA, LMOD2, LRRC10, LZTR1, MAP2K1, MAP2K2, MAP3K8, MIPEP , MLYCD, MRPL3, MRPL44, MRPS22, MT-ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MT-RNR1, MT-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH,   MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY, MTO1, MYBPC3, MYBPHL, MYH6, MYH7, MYL2, MYL3, MYL4, MYO18B, MYOT, MYPN, MYRF, NDUFAF2, NEXN, NF1, NKX2-5, NONO, NOS1AP, NRAP, NRAS, NUP155, PARS2, PCCA, PCCB , PKP2, PLEC, PLEKHM2, PLN, PNPLA2, POMT1, PPA2 PPCS, PPP1CB, PRDM16, PRKAG2, PTPN11, QRSL1, RAF1, RASA2, RBCK1, RBM20, RIT1, RMND1 , RRAS, RYR2, SALL4, SCN10A, SCN1B, SCN3B, SCN5A, SCNN1B, SCNN1G, SCO1, SCO2, SDHA , SELENON , SGCA, SGCB, SGCD, SGCG, SHOC2, SLC22A5, SLC25A20, SLC25A3, SLC25A4, SMCHD1, SOS1, SOS2, SPEG, SPRED1, STAG2, TAB2, TAZ, TBX20 , TBX5, TCAP, TECRL, TGFB3, TMEM43, TMEM70, TNNC1, TNNI3, TNNI3K, TNNT2, TOR1AIP1, TPM1, TRDN, TRIM32, TRPM4, TSFM , TTN, TTR, VARS2, VCL, VCP, VPS13A, XK

10. Dilate Kardiyomiyopati (DCM) Paneli Genleri;

- ✓ ABCC6, ABCC9, ACTA1, ACTC1, ACTN2, ALMS1, ALPK3, APOA1, BAG3, CASZ1, CHRM2, DES, DMD, DOLK, DPM3, DSC2, DSG2, DSP, DYSF, EEF1A2, EMD, EPG5, ETFA, ETFB, ETFDH, FBXO32, FHOD3, FKTN, FLNC , FOXD4 , GATA4 , GATA6, GATC , GBE1, GLB1, GSK3B, HAND1, HCN4, ILK, JPH2, JUP, KLHL24, LAMP2, LDB3, LEMD2, LMNA, LMOD2, LRRC10, MLYCD, MT-ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MT-RNR1, MT-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY, MYBPC3, MYBPHL, MYH6, MYH7, MYL4, NKX2-5, NRAP, PCCA, PCCB, PKP2, PLEKHM2, PLN, PPCS, PRDM16, QRSL1, RAF1, RBCK1, RBM20, RMND1, SCN5A, SPEG, TAB2, TAZ, TBX20, TBX5, TCAP, TMEM43, TNNC1, TNNI3, TNNI3K, TNNT2, TOR1AIP1, TPM1, TTN, TTR, VCL, VPS13A

11. Ehlers-Danlos Sendromu Paneli Genleri;

- ✓ ABCC6 , ADAMTS2, ALDH18A1, ATP6V0A2, ATP7A, B4GALT7, BGN, CBS, CHST14, COL11A1, COL1A1, COL1A2, COL2A1, COL3A1, COL5A1, COL5A2, DSE , EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLNA, PLOD1, PYCR1, SLC39A13, SMAD3, TGFB2, TGFBR1, TGFBR2, ZNF469

12. Kalıtsal Hemorajik Telanjiektazi (HHT) Paneli Genleri;

- ✓ ACVRL1, ENG, EPHB4, GDF2, RASA1, SMAD4

13. Heterotaksi ve Situs İnversus Paneli Genleri;

✓ ACVR2B, ANKS6, ARMC4, C21ORF59, CCDC103, CCDC114, CCDC151, CCDC39, CCDC40, CFAP53, DNAAF1, DNAAF2, DNAAF3, DNAAF5, DNAH11 , DNAH5, DNAI1, DNAI2, DNAL1, DYX1C1, FOXH1, GDF1, INVS, LEFTY2 , LRRC6, MMP21, NODAL, PIH1D3, PITRM1, PKD1L1, SPAG1, TTC25, ZIC3, ZMYND10

14. Hiperlipidemi Çekirdek Paneli Genleri;

✓ APOB, LDLR, LDLRAP1, PCSK9

15. Hiperlipidemi Paneli Genleri;

✓ ABCA1, ABCG5, ABCG8, ALMS1 , APOA1, APOA5, APOB, APOC2, APOC3, APOE, CREB3L3, GPIHBP1, LDLR, LDLRAP1, LIPA, LMF1, LPL, PCSK9

16. Hipertrofik Kardiyomiyopati (HKMP) Paneli Genleri;

✓ ABCC9, ACAD9, ACADVL, ACTA1, ACTC1, ACTN2, AGK , AGL, ALPK3, APOA1, BAG3, BRAF , CBL, COX15, CSRP3, ELAC2, EPG5, FBXL4, FHL1 , FHOD3, FLNC , FXN , GAA, GLA, GSK3B, HRAS, JPH2, KLHL24, LAMP2, MIPEP , MT-ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB,   MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MT-RNR1, MT-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG,   MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY, MYBPC3, MYH7, MYL2, MYL3, NDUFAF2, PLN, PRKAG2, PTPN11, RAF1, RIT1, SLC25A4, SOS1, TNNC1, TNNI3, TNNT2, TPM1, TTR

17. Sol Ventriküler Sıkıştırılmamış Kardiyomiyopati (LVNC) Paneli Genleri;

- ✓ ABCC9, BAG3, CTNNA3, DES, DMD, DSC2, DSG2, DSP, DTNA, EMD, FBXO32, FLNC , HCN4, JPH2, JUP, LAMP2, LMNA, MIPEP , MYBPC3, MYH6, MYH7, PKP2 , PLEKHM2, PLN, RAF1, RBM20, RYR2, SCN5A, TCAP, TNNT2, TPM1, TTN , VCL

18. Liddle Sendromu Paneli Genleri;

- ✓ SCNN1B, SCNN1G

19. Uzun QT Sendromu (LQTS) Paneli Genleri;

- ✓ AKAP9, ANK2, CACNA1C, CALM1 , CALM2, CALM3, CAV3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ5, KCNQ1, NOS1AP, SCN5A, TECRL

20. Marfan Sendromu Paneli Genleri;

- ✓ ABL1, ADAMTS10, ADAMTS17, ADAMTSL4, B3GAT3 , BGN, CBS, COL11A1, COL11A2, COL1A1, COL1A2, COL2A1, COL3A1, COL5A1, COL5A2, DLG4, EFEMP2, FBN1, FBN2, LOX, MAT2A , MED12, PLOD1, SKI, SLC2A10, SMAD3, SMAD6, TGFB2, TGFB3, TGFBR1, TGFBR2, UPF3B, VCAN, ZDHHC9

21. Noonan Sendromu Paneli Genleri;

- ✓ ACTB , ACTG1, BRAF, CBL, CCNK, CDC42 , EPHB4, FGD1, HRAS, KAT6B, KRAS , LZTR1, MAP2K1, MAP2K2, MAP3K8, MRAS, NF1 , NF2, NRAS, NSUN2, PPP1CB, PTPN11, RAF1, RASA1, RASA2, RIT1, RRAS, SASH1, SHOC2, SMARCB1, SOS1, SOS2, SPRED1, STAMBP, SYNGAP1

22. Pulmoner Hipertansiyon (PAH) Paneli Genleri;

✓ ABCC8, ACVRL1, AQP1, ATP13A3, BMPR1B, BMPR2, CAV1, EIF2AK4, ENG, FOXF1, GDF2, KCNA5, KCNK3, KLF2, NFU1, NOTCH3, RASA1, SARS2, SMAD4, SMAD9, SOX17, STRA6, TBX4

23. Kısa QT Sendromu (SQTS) Paneli Genleri;

✓ CACNA1C , CACNB2, KCNH2, KCNJ2, KCNQ1