

MALFORMASYONLAR VE GENETİK POLİKLİNİĞİ

Vücuttaki kemik ve kıkırdaklarda meydana gelen değişimler sonucu iskelet sistemi yapısının bozulması, ciltte meydana gelen lekelenmeler, beynin aşırı büyümesi ya da küçülmesi, atipik yüz görünümü, lipid metabolizması bozuklukları gibi vücutta bulunan bütün organların kan, damar, lipid, kemik ve kıkırdak yapısının anormal gelişmesine bağlı olarak meydana gelen değişimlerin incelenmesi için yapılan çalışmaların olduğu panellerdir.

1. 3-M Sendromu / Primordial Cücelik Paneli Genleri;

✓ ATR, BCS1L, CCDC8, CDC45, CDC6, CDT1, CENPJ, CEP152, CEP63, CUL7, LARP7, NOTCH2, OBSL1, ORC1, ORC4, ORC6, PCNT, POC1A, RBBP8, RNU4ATAC, RTTN, SRCAP, TRIM37, XRCC4

2. Adams-Oliver Sendromu Paneli Genleri;

✓ ARHGAP31, DLL4, DOCK6, EOGT, KCTD1, NOTCH1, RBPJ, UBR1

3. Amelogenesis Imperfekta ve Dentinogenesis Imperfekta Paneli Genleri;

✓ AMELX, C4ORF26, DLX3, DSPP, ENAM, FAM20A, FAM83H, GPR68, ITGB6, KLK4, LAMB3, LTBP3, MMP20, SLC24A4, WDR72, WNT10B

**4. Artrogripozis Paneli
Genleri;**

- ✓ ACTA1, ADGRG6, AGRN, BIN1, CACNA1E, CASK, CFL2, CHAT, CHRNA1, CHRN1, CHRND, CHRNE, CHNG, CHST14, CHUK, CNTNAP1, COL6A2, COLQ, DHCR24, DOK7, DPAGT1, ECEL1, EGR2, ERBB3, ERCC5, ERCC6, EXOSC3, FBN2, FHL1, FKBP10, FKTN, FLVCR2, GBA, GBE1, GFPT1, GLDN, GLE1, KAT6B, KIAA1109, KLHL40, LGI4, LMNA, MPZ, MTM1, MUSK, MYBPC1, MYH2, MYH3, MYH8, NALCN, NEB, PIEZO2, PLOD2, PMM2, PPP3CA, RAPS, RARS2, RIPK4, SCO2, SELENON, SMN1, SMN2, TGFB3, TK2, TNNI2, TNNT1, TNNT3, TPM2, TPM3, TRPV4, TSEN2, TSEN54, UBA1, VIPAS39, VPS33B, VRK1, ZBTB42, ZC4H2

**5. Brakidaktili/
Sindaktili Paneli
Genleri;**

- ✓ BMP2, BMPR1B, CHSY1, DHCR7, ESCO2, FAM58A, GDF5, GNAS, HOXA13, HOXD13, IHH, MYCN, NOG, PDE3A, PDE4D, PTDSS1, PTHLH, RECQL4, ROR2, SOX9, TP63

**6. Serebral Kavernöz
Malformasyon Paneli
Genleri;**

- ✓ CCM2, KRIT1, PDCD10, RASA1

**7. Kondrodisplazi
Punktata Paneli
Genleri;**

- ✓ AGPS, ARSE, EBP, GNPAT, LBR, NSDHL, PEX14, PEX19, PEX7

**8. Yarık Dudak/Damak
ve İlişkili Sendromlar
Paneli Genleri;**

- ✓ ARHGAP29, COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, COL9A3, CTNND1, FOXE1, GRHL3, IRF6, KDM6A, KMT2D, MSX1, SATB2, SPECC1L, TBX2, TBX22, TGDS, TP63, TXNL4A, ZSWIM6

**9. Kapsamlı
Büyüme
Bozuklukları /
İskelet
Displazileri ve
Bozuklukları
Paneli Genleri;**

- ✓ ACAN, ACP5, ACTB, ACTG1, ACVR1, ADAMTS10, ADAMTS17, ADAMTSL2, AGPS, AIFM1, AKT1, ALPL, ALX3, ALX4, AMER1, AMMECR1, ANKH, ANKRD11, ANO5, ARCN1, ARHGAP31, ARID1A, ARID1B, ARSB, ARSE, ATP6V0A2, ATR, B3GALT6, B3GAT3, B4GALT7, BCS1L, BGN, BHLHA9, BMP1, BMP2, BMPER, BMPR1B, BRAF, BRCA2, BRIP1, CA2, CANT1, CASR, CBL, CCDC47, CCDC8, CDC42, CDC45, CDC6, CDH11, CDKN1C, CDT1, CENPJ, CEP152, CEP63, CHST14, CHST3, CHSY1, CKAP2L, CLCN5, CLCN7, COL10A1, COL11A1, COL11A2, COL1A1, COL1A2, COL27A1, COL2A1, COL3A1, COL5A1, COL5A2, COL9A1, COL9A2, COL9A3, COMP, CREB3L1, CREBBP, CRLF1, CRTAP, CSPP1, CTSK, CUL7, CYP27B1, DDR2, DHCR24, DHCR7, DHODH, DLL3, DLL4, DLX3, DLX5, DMP1, DOCK6, DONSON, DVL1, DYM, DYNC2H1, EBP, EFNB1, EFTUD2, EIF2AK3, ENAM, ENPP1, EOGT, EP300, ERCC4, ESCO2, EVC, EVC2, EXT1, EXT2, EXTL3, EZH2, FAM111A, FAM20A, FAM20C, FAM46A, FAM58A, FAM83H, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FBN1, FBN2, FGD1, FGF10, FGF23, FGFR1, FGFR2, FGFR3, FKBP10, FLNA, FLNB, FN1, GALNT3, GDF5, GH1, GHR, GHRHR, GHSR, GJA1, GLI2, GLI3, GNAS, GNPAT, GPC6, HDAC8, HESX1, HOXA13, HOXD13, HRAS, HSPG2, IDS, IDUA, IFITM5, IFT122, IFT140, IFT172, IFT43, IFT52, IFT80, IGF1, IGF1R, IGFALS, IHH, IMPAD1, INPPL1, INSR, IRS1, KAT6B, KIF22, KIF7, KMT2A, KRAS, KYNU, LARP7, LBR, LEMD3, LFNG, LHX3, LHX4, LIFR, LMNA, LMX1B, LONP1, LRP4, LRP5, LTBP2, LTBP3, LZTR1, MAFB, MAP2K1, MAP2K2, MAP3K7, MATN3, MBTPS2, MESP2, MGP, MMP13, MMP2, MMP9, MSX2, MYCN, MYO18B, NANS, NEK1, NF1, NFIX, NIPBL, NKX3_2, NOG, NOTCH1, NOTCH2, NPR2, NRAS, NSD1, NSDHL, OBSL1, ORC1, ORC4, ORC6, OSGEP, OSTM1, OTX2, P3H1, PALB2, PAPSS2, PCNT, PCYT1A, PDE3A, PDE4D, PEX14, PEX19, PEX7, PGM3, PHEX, PIK3CA, PISD, PITX1, PITX2, PLOD2, PLS3, POC1A, POLA1, POLR1C, POLR1D, POP1, POR, POU1F1, PPIB, PPP3CA, PRKAR1A, PRMT7, PROP1, PTDSS1, PTH1R, PTHLH, PTPN11, PUF60, PYCR1, RAB33B, RAD21, RAD51C, RAF1, RALA, RASA2, RBBP8, RBM8A, RBPJ, RECQL4, RIT1, RMRP, RNU4ATAC, ROR2, RRAS, RSPRY1, RTTN, RUNX2, SALL1, SALL4, SBDS, SEC24D, SERPINF1, SERPINH1, SETBP1, SF3B4, SGMS2, SH3BP2, SH3PXD2B, SHOC2, SHOX, SKI, SLC26A2, SLC29A3, SLC34A3, SLC35D1, SLC39A13, SLCO2A1, SLX4, SMAD3, SMAD4, SMARCA2, SMARCAL1, SMARCE1, SMC1A, SMC3, SNX10, SOS1, SOST, SOX11, SOX2, SOX3, SOX9, SP7, SPARC, SRCAP, STAG2, STAMBP, STAT5B, TALDO1, TBX15, TBX19, TBX2, TBX3, TBX4, TBX5, TBX6, TCF12, TCIRG1, TCOF1, TCTEX1D2, TCTN3, TGFB1, TGFB2, TGFB3, TGFB1, TGFB2, TMEM38B, TNFRSF11A, TNFRSF11B, TNFSF11, TONSL, TOP3A, TP63, TRAPPC2, TRIM37, TRIP11, TRMT10A, TRPS1, TRPV4, TTC21B, TWIST1, TYROBP, VDR, VIPAS39, WDR19, WDR34, WDR35, WDR60, WISP3, WNT1, WNT5A, WNT7A, XRCC2, XRCC4, XYLT1, XYLT2

10. Kapsamlı Kısa Boy Sendromu Paneli Genleri;

- ✓ ACTB, ACTG1, AMMECR1, ARCN1, ATR, B3GAT3, BCS1L, BRAF, CBL, CCDC8, CDC45, CDC6, CDT1, CENPJ, CEP152, CEP63, COL27A1, CREBBP, CUL7, DHCR7, DONSON, EP300, FGD1, FGFR3, FN1, GH1, GHR, GHRHR, GHSR, GLI2, GNAS, HDAC8, HESX1, HRAS, IDUA, IGF1, IGF1R, IGFALS, INSR, IRS1, KRAS, LARP7, LFNG, LHX3, LHX4, LZTR1, MAP2K1, MAP2K2, NIPBL, NOTCH2, NRAS, OBSL1, ORC1, ORC4, ORC6, OSGEP, OTX2, PCNT, PISD, PITX2, POC1A, POP1, POU1F1, PPP3CA, PRMT7, PROP1, PTPN11, PUF60, RAD21, RAF1, RALA, RASA2, RBBP8, RIT1, RNU4ATAC, RRAS, RTTN, SGMS2, SHOC2, SHOX, SMARCA2, SMARCE1, SMC1A, SMC3, SOS1, SOX11, SOX2, SOX3, SRCAP, STAT5B, TALDO1, TBX19, TBX2, TBX3, TOP3A, TRIM37, TRMT10A, XRCC4

11. Kapsamlı İskelet Displazileri ve Bozuklukları Paneli Genleri;

- ✓ ACAN, ACP5, ACVR1, ADAMTS10, ADAMTS17, ADAMTSL2, AGPS, AIFM1, AKT1, ALPL, ALX3, ALX4, AMER1, ANKH, ANKRD11, ANO5, ARHGAP31, ARSB, ARSE, ATP6V0A2, B3GALT6, B3GAT3, B4GALT7, BGN, BHLHA9, BMP1, BMP2, BMPER, BMPR1B, CA2, CANT1, CASR, CDC45, CDC6, CDKN1C, CDT1, CHST14, CHST3, CHSY1, CKAP2L, CLCN5, CLCN7, COL10A1, COL11A1, COL11A2, COL1A1, COL1A2, COL27A1, COL2A1, COL9A1, COL9A2, COL9A3, COMP, CREBBP, CRLF1, CRTAP, CSPP1, CTSK, CUL7, CYP27B1, DDR2, DHCR24, DLL3, DLL4, DLX3, DMP1, DOCK6, DVL1, DYM, DYNC2H1, EBP, EFN1, EFTUD2, EIF2AK3, ENAM, ENPP1, EOGT, EP300, ESCO2, EVC, EVC2, EXT1, EXT2, EXTL3, EZH2, FAM111A, FAM20A, FAM20C, FAM83H, FANCB, FANCC, FBN1, FBN2, FGF23, FGFR1, FGFR2, FGFR3, FKBP10, FLNA, FLNB, GALNT3, GDF5, GJA1, GLI3, GNAS, GNPAT, GPC6, HDAC8, HOXA13, HOXD13, HSPG2, IDS, IDUA, IFT122, IFT140, IFT172, IFT43, IFT80, IHH, IMPAD1, INPPL1, KAT6B, KIF22, KIF7, KMT2A, LBR, LEMD3, LIFR, LMNA, LMX1B, LONP1, LRP4, LRP5, LTBP2, LTBP3, MAFB, MATN3, MESP2, MGP, MMP13, MMP2, MMP9, MSX2, MYCN, MYO18B, NANS, NEK1, NF1, NFIX, NIPBL, NKX3-2, NOG, NOTCH2, NPR2, NSD1, NSDHL, OBSL1, ORC1, ORC4, ORC6, OSTM1, P3H1, PAPSS2, PCNT, PCYT1A, PDE4D, PEX7, PGM3, PHEX, PIK3CA, PISD, PLOD2, PLS3, POLR1C, POLR1D, POR, PPIB, PRKAR1A, PTDSS1, PTH1R, PTHLH, PTPN11, PYCR1, RAB33B, RAD21, RBPJ, RECQL4, RMRP, RNU4ATAC, ROR2, RUNX2, SBDS, SERPINF1, SERPINH1, SETBP1, SF3B4, SH3BP2, SH3PXD2B, SHOX, SKI, SLC26A2, SLC29A3, SLC34A3, SLC35D1, SLC39A13, SLCO2A1, SMAD3, SMAD4, SMARCAL1, SMC1A, SMC3, SNX10, SOST, SOX9, SP7, STAMBP, TBX15, TBX3, TBX4, TBX6, TCF12, TCIRG1, TCOF1, TCTN3, TGFB1, TGFB2, TGFB3, TGFB1R1, TGFB2R2, TMEM38B, TNFRSF11A, TNFRSF11B, TNFSF11, TP63, TRAPPC2, TRIP11, TRPS1, TRPV4, TTC21B, TWIST1, TYROBP, VDR, VIPAS39, WDR19, WDR34, WDR35, WDR60, WISP3, WNT1, WNT5A, XYLT1

<input type="checkbox"/>	12. Cornelia de Lange Sendromu Paneli Genleri;	✓ AFF4, HDAC8, NIPBL, RAD21, SMC1A, SMC3
<input type="checkbox"/>	13. Kraniosinostoz Paneli Genleri;	✓ ALPL, ALX3, ALX4, BMP4, CDC45, EDNRB, EFNB1, ERF, ESCO2, FGFR1, FGFR2, FGFR3, FLNB, FREM1, GDF5, GLI3, IFT122, IFT140, IL11RA, MASP1, MEGF8, MSX2, NOG, PAX3, POR, RAB23, RECQL4, SKI, SOX10, SPECC1L, TCF12, TGFBR1, TGFBR2, TWIST1, TWIST2, WDR19, WDR35, ZIC1
<input type="checkbox"/>	14. Ekzostoz ve İlgili Bozukluklar Paneli Genleri;	✓ EXT1, EXT2, PTPN11
<input type="checkbox"/>	15. Yüz Disostoza ve İlgili Bozukluklar Paneli Genleri;	✓ ALPL, ALX3, ALX4, CREBBP, DHODH, DLL3, EFNB1, EFTUD2, EHMT1, EP300, EVC, EVC2, HDAC8, HSPG2, LIFR, MYCN, NIPBL, POLR1A, POLR1C, POLR1D, SF3B4, SMC1A, SMC3, SRCAP, TCOF1, TWIST1, UBE2A
<input type="checkbox"/>	16. Gastrointestinal Atrezi Paneli Genleri;	✓ CDK9, CHD7, CLMP, DHCR7, EFTUD2, FANCB, FANCC, GLI3, MID1, MNX1, MYCN, PTF1A, RFX6, SOX2, TTC7A
<input type="checkbox"/>	17. Heterotaksi ve Situs Inversus 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



18. Hirschsprung Hastalığı Paneli Genleri;

- ✓ BDNF, CELSR3, EDN3, EDNRB, KIF1BP, LICAM, MITF, NRG1, NRTN, PAX3, PHOX2B, RET, RMRP, SOX10, ZEB2



19. Holoprozensefali Paneli Genleri;

- ✓ CDON, FGF8, FGFR1, FOXH1, GLI2, GLI3, NODAL, PTCH1, SHH, SIX3, TGIF1, ZIC2



20. Kabuki Sendromu Paneli Genleri;

- ✓ CHD7, EYA1, FLNB, HNRNPK, IRF6, KDM1A, KDM6A, KMT2D, SIX5



21. Ekstremitte Bozuklukları Paneli Genleri;

- ✓ ARHGAP31, ARID1A, ARID1B, ATR, BHLHA9, BRCA2, BRIP1, DHODH, DLL4, DLX5, DOCK6, EOGT, ERCC4, ESCO2, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FGF10, GDF5, HDAC8, KYNU, NIPBL, NOTCH1, NSDHL, PALB2, PITX1, PUF60, RAD21, RAD51C, RBM8A, RBPJ, RECQL4, SALL1, SALL4, SF3B4, SLX4, SMC1A, SMC3, TBX3, TBX5, TP63, WNT7A, XRCC2



22. Lisensefali Paneli Genleri;

- ✓ ACTB, ACTG1, ARX, ATP6V0A2, CIT, CRADD, DCX, FKTN, ISPD, KATNB1, KIAA1109, LAMB1, LARGE, MACF1, PAFAH1B1, POMGNT2, POMT1, RELN, TMTC3, TUBA1A, TUBB2B, TUBG1, VLDLR, YWHAE

23. Lenfatik Malformasyonlar ve İlişkili Hastalıklar Paneli Genleri;

- ✓ ADAMTS3, CCBE1, FAT4, FLT4, FOXC2, GATA2, GJC2, KIF11, PIEZO1, PIK3CA, RASA1, SOX18

24. Makrosefali / overgrowth ilişkili sendromlar Paneli Genleri;

- ✓ AKT1, AKT3, ASPA, ASXL2, BRWD3, CCND2, CDKN1C, CHD8, CUL4B, DHCR24, DIS3L2, DNMT3A, EED, EIF2B5, EZH2, GFAP, GLI3, GPC3, GPSM2, GRIA3, HEPACAM, HUWE1, KDM1A, KIAA0196, KIF7, KPTN, L1CAM, MED12, MLC1, MPDZ, NFIB, NFIX, NSD1, OFD1, PIGA, PIK3CA, PIK3R2, PTCH1, PTEN, RAB39B, RNF135, SETD2, SYN1, TMEM94, TSC1, TSC2, UPF3B, ZBTB20

25. Meier-Gorlin Sendromu Paneli Genleri;

- ✓ CDC45, CDC6, CDT1, ORC1, ORC4, ORC6

26. Metafizyal Displazi Paneli Genleri;

- ✓ ANKH, CDKN1C, COL10A1, FGFR3, FLNA, MMP13, MMP9, PISD, PTH1R, RMRP, RUNX2, SBDS

**27. Mikrosefali ve
Pontoserebellar
Hipoplazi Paneli
Genleri;**

- ✓ AKT3, AMPD2, ASPM, ASXL1, ASXL3, ATR, CASK, CCDC47, CDK5RAP2, CENPF, CENPJ, CEP152, CEP63, CSNK2A1, DONSON, DYNC1H1, DYRK1A, EFTUD2, EXOSC3, GFM1, GPT2, KANSL1, KATNB1, KIF11, LIG4, MBD5, MCPH1, MED17, MFSD2A, MIPEP, MRE11A, MYCN, MYO18B, NCAPD3, NDE1, NHEJ1, OPHN1, PAFAH1B1, PCDH12, PCLO, PCNT, PHGDH, PLK4, PNKP, POMT1, PQBP1, QARS, RARS2, RTTN, SEPSECS, SLC1A4, SMARCA2, SMARCE1, SOX11, STAG2, STAMBIP, STIL, TBC1D20, TBC1D23, THOC6, TMTC3, TOE1, TOP3A, TRMT10A, TSEN2, TSEN54, TUBB, TUBB2B, TUBGCP4, TUBGCP6, UBE3B, VARS, VRK1, WDR62, WDR73, XRCC4, ZNF148, ZNF335

**28. Mikromelik
Displazi Paneli Genleri;**

- ✓ ADAMTS10, ADAMTSL2, BMPR1B, DVL1, EXT1, FBN1, FGFR3, GDF5, GNAS, IFT122, IFT140, IHH, INPPL1, LIFR, LTBP2, NPR2, PRKAR1A, ROR2, SHOX, SLC35D1, SMAD4, SOX9, TRIP11, TRPS1, WDR19, WDR35, WNT5A

**29. Nörofibromatozis
Paneli Genleri;**

- ✓ KIT, KITLG, LZTR1, NF1, NF2, PTPN11, RAF1, SMARCB1, SPRED1

**30. Nöronal Göç
Bozukluğu Paneli
Genleri;**

- ✓ ACTB, ACTG1, ADGRG1, AKT3, ARFGEF2, ARX, ATP6V0A2, B3GALNT2, COL4A1, COL4A2, DCX, DYNC1H1, EMX2, FAT4, FH, FKTN, FLNA, FLVCR2, GMPPB, GPSM2, ISPD, KATNB1, KIF1BP, KIF7, L1CAM, LAMA2, LAMB1, LAMC3, LARGE, MACF1, MED12, MEF2C, MPDZ, NDE1, NSDHL, OCLN, PAFAH1B1, PHGDH, PIK3CA, PIK3R2, POMGNT2, POMT1, RAB18, RAB3GAP1, RAB3GAP2, RELN, RTTN, SEPSECS, SRPX2, TMEM5, TUBA1A, TUBA8, TUBB2A, TUBB2B, TUBB3, TUBG1, VLDLR, WDR62, YWHAE

**31. Osteogenesis
İmperfekta Paneli
Genleri;**

- ✓ ALPL, ARCN1, B3GAT3, B4GALT7, BMP1, CLCN5, COL1A1, COL1A2, CREB3L1, CRTAP, FAM46A, FGF23, FKBP10, IFITM5, LRP5, MBTPS2, P3H1, PHEX, PLOD2, PLS3, PPIB, SEC24D, SERPINF1, SERPINH1, SGMS2, SLC34A3, SP7, SPARC, TMEM38B, WNT1

**32. Osteopetrosis ve
Yoğun Kemik
Displazisi Paneli
Genleri;**

- ✓ AMER1, ANKH, CA2, CLCN7, COL1A1, CTSK, DLX3, FAM20C, GJA1, LEMD3, LRP4, LRP5, OSTM1, PTDSS1, PTH1R, SLC29A3, SLCO2A1, SNX10, SOST, TCIRG1, TGFB1, TNFRSF11A, TNFRSF11B, TNFSF11, TYROBP

**33. Polimikrogrfi Paneli
Genleri;**

- ✓ ADGRG1, AKT3, FH, GPSM2, KIF1BP, LAMC3, NDE1, NSDHL, OCLN, PI4KA, RAB18, SNAP29, SRPX2, TBC1D20, TUBA1A, TUBA8, TUBB2A, TUBB2B, TUBB3, WDR62

- 34. Seckel Sendromu Paneli Genleri;**
 - ✓ ATR, CENPJ, CEP152, CEP63, PCNT, RBBP8
- 35. Septo-Optik Displazi Paneli Genleri;**
 - ✓ HESX1, OTX2, PAX6, SOX2
- 36. Shortrib/ Asphyxiating Thoracic Displazi Paneli Genleri;**
 - ✓ CSPP1, DYNC2H1, EVC, EVC2, GLI2, IFT122, IFT140, IFT172, IFT52, IFT80, NEK1, TCTEX1D2, TCTN3, TTC21B, WDR19, WDR34, WDR35, WDR60
- 37. Anormal Mineralizasyon-İskelet Displazisi Paneli Genleri;**
 - ✓ ALPL, ANKH, B4GALT7, CASR, CLCN5, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, CRTAP, CYP27B1, ENPP1, FBN1, FGF23, FKBP10, GALNT3, MGP, P3H1, PHEX, PLOD2, PLS3, PPIB, PTDSS1, SERPINF1, SGMS2, SLC34A3, SLC39A13, SNX10, SOX9, TNFRSF11A, TNFRSF11B, VDR, XYLT2

**38. İskelet Displazileri
Çekirdek Paneli
Genleri;**

- ✓ ACAN, ACP5, ADAMTS10, ADAMTSL2, AGPS, ALPL, ANKH, ARSE, B3GALT6, BMP1, BMPR1B, CA2, CANT1, CDC6, CDKN1C, CDT1, CHST3, CLCN7, COL10A1, COL11A1, COL11A2, COL1A1, COL1A2, COL2A1, COL9A1, COL9A2, COL9A3, COMP, CRTAP, CSPP1, CTSK, CUL7, CYP27B1, DHCR24, DLL3, DVLI, DYM, DYNC2H1, EBP, EIF2AK3, ENPP1, ESCO2, EVC, EVC2, FAM20C, FGF23, FGFR1, FGFR2, FGFR3, FKBP10, FLNA, FLNB, GDF5, GNPAT, HSPG2, IFT140, IFT172, IFT80, IHH, INPPL1, KAT6B, LBR, LIFR, LMX1B, LRP5, LTBP2, MATN3, MMP9, MYO18B, NEK1, NPR2, OBSL1, ORC1, ORC4, ORC6, P3H1, PAPSS2, PCNT, PEX7, PHEX, PISD, PLOD2, PLS3, PPIB, PTH1R, RMRP, RNU4ATAC, ROR2, RUNX2, SBDS, SERPINF1, SERPINH1, SHOX, SLC26A2, SLC34A3, SLC39A13, SMAD4, SMARCAL1, SOX9, TCIRG1, TGFB1, TNFRSF11A, TNFRSF11B, TRAPPC2, TRIP11, TRPV4, TTC21B, VDR, WDR19, WDR35, WISP3, WNT5A, XYLT1

**39. Spondilometafizyal /
Spondiloepi (meta) -
fizyal Displazi Paneli
Genleri;**

- ✓ ACAN, ACP5, B3GALT6, BGN, CANT1, CHST3, COL11A1, COL11A2, COL2A1, DDR2, DYM, EIF2AK3, HSPG2, INPPL1, KIF22, LONP1, MATN3, MMP13, NKX3-2, PAPSS2, PCYT1A, PISD, RAB33B, RMRP, RSPRY1, SLC39A13, SMARCAL1, TRAPPC2, TRPV4, WISP3

**40. Vasküler
Malformasyonlar
Paneli Genleri;**

- ✓ ACVRL1, CCM2, ELMO2, ENG, GLMN, KRIT1, PDCD10, PIK3CA, PTEN, RASA1, SMAD4, SOX18, STAMBP, TEK