

KULAK BURUN BOĞAZ VE GENETİK POLİKLİNİĞİ

İşitme kaybı ve bu kayıpları da içeren kulak malformasyonları, aşırı genişlemiş kapiller ve venüllerin neden olduğu kanamalar, tiroid beziyle ilişkili doğuştan gelen işitme kaybı, iç kulak anomalisinden kaynaklanan işitme azlığı ya da kaybı, işitsel pigmenterlik gibi kulak, burun, boğaz bölümünü ilgilendiren ve herediter özellik taşıyan bu durumlara cevap bulmak ve tedaviyi kolaylaştırıp yenidoğan ünitelerinde ve prenatal tanıda erken teşhis oluşturabilmek için yapılan çalışmaların olduğu panellerdir.

1. Alport Sendromu Paneli Genleri;

✓ CD151, COL4A3, COL4A4, COL4A5, COL4A6, MYH9

2. Branchio-Oto-Renal (BOR) Sendrom Paneli Genleri;

✓ EYA1, SIX1, SIX5, TFAP2A

3. Kapsamlı İşitme Kaybı ve Sağırılık Paneli Genleri;

- ✓ ABHD12, ACTG1 , ADCY1, ADGRV1, AIFM1, ALMS1 , ANKH, ARSG, ATP2B2, ATP6V1B1, ATP6V1B2, BCS1L, BDP1 , BSND, BTD, C10ORF2, CABP2, CACNA1D, CATSPER2, CCDC50, CD151, CD164, CDC14A, CDC42 , CDH23, CDK9, CDKN1C, CEACAM16, CEP250, CEP78, CHD7, CHSY1, CIB2, CLDN14, CLIC5, CLPP, CLRN1, COCH, COL11A1, COL11A2, COL2A1, COL4A3, COL4A4, COL4A5, COL4A6, COL9A1, COL9A2, COL9A3, CRYM, DCAF17, DCDC2, DFNA5, DFNB31, DFNB59, DIABLO, DIAPH1, DIAPH3, DLX5, DMXL2, DNMT1, DSPP, EDN3, EDNRB, EIF3F, ELMOD3, EPS8, EPS8L2, ESPN , ESRRB, EYA1, EYA4, FAM136A , FAM65B, FDXR, FGF3, FGFR3, FITM2, FOXI1, GATA3, GIPC3, GJA1, GJB2, GJB3, GJB6, GPSM2, GRHL2, GRXCR1, GRXCR2, HARS, HARS2, HGF, HOMER2, HOXB1, HSD17B4, ILDR1, KARS, KCNE1, KCNJ10, KCNQ1, KCNQ4, KIT, LARS2, LHFPL5, LMX1A, LOXHD1, LRP2, LRTOMT, MAN2B1, MANBA, MARVELD2, MET, MGP, MIR96, MITF, MPZL2, MSRB3, 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, MYH14, MYH9, MYO15A, MYO3A, MYO6, MYO7A, NARS2, NDP, NLRP3, OSBPL2, OTOA, OTOF, OTOG, OTOGL, P2RX2, PAX3, PCDH15, PDE1C, PDZD7 , PEX1, PEX26, PEX6, PISD, PNPT1 , POLR1C , POLR1D, POU3F4, POU4F3, PRPS1 , RDX , RMND1 , RPS6KA3, S1PR2, SALL1 , SALL4, SEMA3E, SERPINB6, SIX1, SIX5, SLC17A8, SLC19A2, SLC22A4, SLC26A4, SLC26A5, SLC29A3, SLC33A1 , SLC52A2, SLC52A3, SLITRK6, SMAD4, SMPX, SNAI2, SOX10, SPATA5, STAG2, STRC , SUCLA2, SUCLG1, SYNE4, SYT2 , TBC1D24, TBL1X, TCOF1, TECTA, TFAP2A, TIMM8A , TJP2, TMC1, TMEM132E, TMIE, TMPRSS3, TNC, TPRN, TRIOBP, TRMU, TSHZ1, TSPEAR, TUBB4B, TYR , USH1C, USH1G, USH2A, VCAN, WBP2, WFS1, XYLT2

**4. Kalıtsal Hemorajik
Telanjektazi (HHT)
Paneli Genleri;**

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

**5. Sendromik Olmayan
İşitme Kaybı Paneli
Genleri;**

✓ ACTG1 , ADCY1, ATP2B2, BDP1 , BSND, CABP2, CCDC50, CD164, CDC14A, CDH23, CEACAM16, CIB2, CLDN14, CLIC5, COCH, COL11A2, COL4A6, CRYM, DCDC2, DFNA5, DFNB31, DFNB59, DIABLO, DIAPH1, DIAPH3, DMXL2, DSPP, ELMOD3, EPS8, EPS8L2, ESPN , ESRRB, EYA4, FAM65B, GIPC3, GJB2, GJB3, GJB6, GPSM2, GRHL2, GRXCR1, GRXCR2, HGF, HOMER2, ILDR1, KARS, KCNQ4, LHFPL5, LMX1A, LOXHD1, LRTOMT, MARVELD2, MET, MIR96, MPZL2, MSRB3, 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, MYH14, MYH9, MYO15A, MYO3A, MYO6, MYO7A, NARS2, OSBPL2, OTOA , OTOF, OTOG, OTOGL, P2RX2, PCDH15, PDE1C, PNPT1, POU3F4, POU4F3, PRPS1, RDX, S1PR2, SERPINB6, SIX1, SLC17A8, SLC22A4, SLC26A4, SLC26A5, SLITRK6, SMPX, STRC, SYNE4, TBC1D24, TECTA, TJP2, TMC1, TMEM132E, TMIE, TMPRSS3, TNC, TPRN , TRIOBP, TSPEAR, USH1C, WBP2, WFS1

**6. Pendred Sendrom
Paneli Genleri;**

✓ FOXI1, KCNJ10, SLC26A4

**7. Stickler Sendromu
Paneli Genleri;**

- ✓ COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, COL9A3, LRP2, VCAN

**8. Sendromik İşitme
Kaybı Paneli Genleri;**

- ✓ ABHD12, ACTG1 , ADGRV1, ALMS1 , ANKH, ARSG, ATP6V1B1, ATP6V1B2, BCS1L, BSND, BTD, C10ORF2, CACNA1D, CD151, CDH23,CDK9, CDKN1C, CEP250, CEP78, CHD7, CHSY1, CIB2, CLPP, CLRN1, COL11A1, COL11A2, COL2A1, COL4A3, COL4A4, COL4A5, COL4A6, COL9A1, COL9A2, COL9A3, DCAF17, DFNB31, DLX5, DNMT1, EDN3, EDNRB, EIF3F, ESPN , EYA1, FDXR, FGF3, FITM2, FOXI1, GATA3, GJA1 , HARS, HARS2, HOXB1, KCNE1, KCNJ10, KCNQ1, KIT, LARS2, LRP2, MAN2B1, MANBA, MGP, MITF, 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, MYH9, MYO7A, NDP, NLRP3, PAX3, PCDH15, PDZD7 , PEX1, PEX26, PEX6, PISD, POLR1C , POLR1D, PRPS, SALL1 , SEMA3E, SIX1, SIX5, SLC19A2, SLC26A4, SLC52A2, SLC52A3, SLITRK6, SMAD4, SNAI2, SOX10, TBL1X, TCOF1, TFAP2A, TIMM8A , TSHZ1, TUBB4B, TYR , USH1C, USH1G, USH2A, WFS1,XYLT2

**9. Usher Sendromu
Paneli Genleri;**

- ✓ ABHD12, ADGRV1, ARSG, CDH23, CEP250, CEP78, CIB2, CLRN1, DFNB31, ESPN , HARS, MYO7A, PCDH15, PDZD7, PEX1, PEX6, PRPS, TUBB4B, USH1C, USH1G, USH2A

**10. Waardenburg
Sendromu Paneli
Genleri;**

- ✓ EDN3, EDNRB, KIT, MITF, PAX3, SNAI2, SOX10