

## OFTALMOLOJİ VE GENETİK POLİKLİNİĞİ

Gece körlüğü, retinada meydana gelen değişimler, göz küresi anomalisi, göz tansiyonu, kornea tabakası bozukluğu, gözdeki bağ dokularının bozukluğu gibi oftalmolojik durumların genetikle bağlantısını ortaya çıkarmak ve prenatal tanıda bu çıkarımları kullanarak tedavi süreci geliştirmek için yapılan çalışmaların olduğu panellerdir.

### 1. Akromatopsi Paneli Genleri;

- ✓ ATF6, CNGA3, CNGB3, GNAT2, PDE6C, PDE6H, RGS9, RGS9BP

### 2. Albinizm Paneli Genleri;

- ✓ AP3B1, BLOC1S3, BLOC1S6, C10ORF11, DTNBP1, GPR143, HPS1, HPS3, HPS4, HPS5, HPS6, LYST, MC1R, MITF, MLPH, MYO5A, OCA2, RAB27A, SLC24A5, SLC38A8, SLC45A2, TYR, TYRP1

### 3. Bardet-Biedl Sendromu Paneli Genleri;

- ✓ ALMS1, ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C8ORF37, CEP19, CEP290, CPE, IFT172, IFT27, LZTFL1, MKKS, MKS1, PNPLA6, SCAPER, SDCCAG8, TMEM67, TRIM32, TTC8, WDPCP

#### 4. Katarakt Paneli Genleri;

- ✓ ABCB6, ADAMTS18, ADAMTSL4, AGK, ALDH18A1, BCOR, BFSP1, BFSP2, CHMP4B, COL11A1, COL18A1, COL2A1, COL4A1, CRYAA, CRYAB, CRYBA1, CRYBA4, CRYBB1, CRYBB2, CRYBB3, CRYGC, CRYGD, CRYGS, CTDPI, CYP27A1, DNMBP, EPHA2, ERCC2, ERCC5, ERCC6, ERCC8, EYA1, FAM126A, FOXE3, FTL, FYCO1, FZD4, GALE, GALK1, GALT, GCNT2, GJA1, GJA3, GJA8, HSF4, LEMD2, LIM2, MAF, MIP, 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, NDP, NF2, NHS, OCRL, OPA3, P3H2, PAX6, PITX3, PXDN, RAB18, RAB3GAP1, RAB3GAP2, RECQL4, SIL1, SIPA1L3, SLC16A12, SLC33A1, TBC1D20, TDRD7, TFAP2A, TMEM70, VIM, VSX2, WFS1, WRN, XYLT2

#### 5. Kon-Rod Distrofi Paneli Genleri;

- ✓ ABCA4, ADAM9, ADAMTS18, AIPL1, ARHGEF18, BEST1, C21ORF2, C8ORF37, CABP4, CACNA1F, CACNA2D4, CDHR1, CEP250, CEP78, CERKL, CLN3, CNGA3, CNGB3, CNNM4, CRB1, CRX, CYP4V2, ELOVL4, GNAT2, GUCA1A, GUCY2D, KCNV2, MERTK, PDE6C, PDE6H, PITPNM3, POC1B, PROM1, PRPH2, RAB28, RAX2, RDH5, RGS9, RGS9BP, RIMS1, RPGR, RPRIP1, SEMA4A, TLL5

#### 6. Konjenital Durağan (Sabit) Gece Körlüğü Paneli Genleri;

- ✓ CABP4, CACNA1F, CACNA2D4, CYP4V2, FRMD7, GNAT1, GNB3, GPR179, GRK1, GRM6, LRIT3, NYX, PDE6B, RDH5, RHO, RLBP1, RPE65, SAG, SLC24A1, TRPM1



**7. Korneal Distrofi  
Paneli Genleri;**

- ✓ CHRDL1, CHST6, COL17A1, COL5A1, COL8A2, CYP4V2, DCN, FOXE3, GJA8, GRHL2, GSN, KERA, KRT12, KRT3, LCAT, LOXHD1, MAF, NLRP3, OVOL2, PIKFYVE, PITX2, PRDM5, SLC4A11, TACSTD2, TCF4, TGFBI, UBIAD1, ZEB1, ZNF469



**8. Ektopia Lentis  
Paneli Genleri;**

- ✓ AASS, ADAMTS10, ADAMTS17, ADAMTSL4, ASPH, BCOR, CBS, COL18A1, FBN1, LTBP2, P3H2, PORCN, SUOX, VSX2



**9. Flecked (Benekli)  
Retina  
Bozuklukları Paneli  
Genleri;**

- ✓ ABCA4, CHM, CYP4V2, ELOVL4, PLA2G5, PROM1, PRPH2, RDH5, RHO, RLBP1, RS1, VPS13B



**10. Glokom Paneli  
Genleri;**

- ✓ CNTNAP2, COL4A1, CYP1B1, FOXC1, FOXE3, LMX1B, LTBP2, MAF, MYOC, OPA1, OPA3, OPTN, PAX6, PITX2, PXDN, TBK1, TEK, TMEM126A, WDR36



**11. Kalıtsal  
Retinoblastoma  
Paneli Genleri;**

- ✓ RB1

**12. Joubert Sendromu Paneli Genleri;**

- ✓ AHI1, ARL13B, ARMC9, B9D1, B9D2, C21ORF2, C5ORF42, CC2D2A, CEP104, CEP120, CEP164, CEP290, CEP41, CSPP1, INPP5E, KIAA0556, KIAA0586, KIAA0753, KIF7, MKS1, NPHP1, NPHP3, OFD1, PDE6D, RPGRIP1L, TCTN1, TCTN2, TCTN3, TMEM107, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TTC21B, ZNF423

**13. Leber Konjenital Amaurosis Paneli Genleri;**

- ✓ AIPL1, ALMS1, BBS4, CABP4, CEP290, CNGA3, CRB1, CRX, CWC27, DTHD1, GUCY2D, IDH3A, IMPDH1, IQCB1, KCNJ13, LCA5, LRAT, MERTK, MYO7A, NMNAT1, RD3, RDH12, RDH5, RPE65, RPGRIP1, SPATA7, TUBB4B, TULP1

**14. Maküler Distrofi Paneli Genleri;**

- ✓ ABCA4, BEST1, C1QTNF5, CDH3, CERKL, CNGB3, CRB1, CRX, CTNNA1, DRAM2, EFEMP1, ELOVL4, IMPG1, IMPG2, KCNV2, MFSD8, NMNAT1, PRDM13, PROM1, PRPH2, RAX2, RDH12, RDH5, RLBP1, RP1L1, RPGR, RS1, TIMP3

**15. Mikroftalmi, Anoftalmi ve Anterior Segment Disgenezi Paneli Genleri;**

- ✓ ABCB6, ADAMTS18, ALDH1A3, BCOR, BMP4, BMP7, CDK9, CHD7, COL4A1, COX7B, CPAMD8, CYP1B1, ERCC2, ERCC5, ERCC6, FOXC1, FOXE3, FOXL2, FRAS1, FREM1, FREM2, GJA1, GRIP1, HCCS, HESX1, HMX1, MAB21L2, MFRP, MITF, NAA10, NDP, OCRL, OTX2, PAX2, PAX6, PITX2, PQBP1, PRSS56, PXDN, RAB18, RAB3GAP1, RAB3GAP2, RARB, RAX, RBP4, SHH, SIPA1L3, SIX3, SIX6, SLC38A8, SMCHD1, SMOC1, SOX2, STRA6, TBC1D20, TENM3, TFAP2A, VPS13B, VSX2, YAP1, ZIC2

**16. Retina Tracker  
Program Paneli  
Genleri;**

- ✓ ABCA4, ABCC6, ABHD12, ACO2, ADAM9, ADAMTS18, ADGRV1, ADIPOR1, AGLB5, AHI1, AIPL1, ALMS1, ARHGEF18, ARL13B, ARL2BP, ARL3, ARL6, ARMC9, ARSG, ATF6, ATOH7, B9D1, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BEST1, C1QTNF5, C21ORF2, C2ORF71, C5ORF42, C8ORF37, CA4, CABP4, CACNA1F, CACNA2D4, CAPN5, CC2D2A, CDH23, CDH3, CDHR1, CEP104, CEP120, CEP164, CEP19, CEP250, CEP290, CEP41, CEP78, CERKL, CHM, CIB2, CISD2, CLN3, CLRN1, CNGA1, CNGA3, CNGB1, CNGB3, CNNM4, COL11A1, COL11A2, COL18A1, COL2A1, COL9A1, COL9A2, COL9A3, CPE, CRB1, CRX, CSPP1, CTC1, CTNNA1, CTNNB1, CWC27, CYP4V2, DFNB31, DHDDS, DHX38, DRAM2, DTHD1, EFEMP1, ELOVL4, EMC1, ESPN, EYS, FAM161A, FDXR, FLVCR1, FRMD7, FZD4, GNAT1, GNAT2, GNB3, GNPTG, GPR179, GRK1, GRM6, GUCA1A, GUCY2D, HARS, HGSNAT, HK1, HMX1, IDH3A, IDH3B, IFT140, IFT172, IFT27, IFT81, IMPDH1, IMPG1, IMPG2, INPP5E, INVS, IQCB1, JAG1, KCNJ13, KCNV2, KIAA0556, KIAA0586, KIAA0753, KIAA1549, KIF11, KIF7, KIZ, KLHL7, LCA5, LRAT, LRIT3, LRP2, LRP5, LZTFL1, MAK, MERTK, MFN2, MFRP, MFSD8, MKKS, MKS1, MMACHC, 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, MTTP, MVK, MYO7A, NDP, NEK2, NMNAT1, NPHP1, NPHP3, NPHP4, NR2E3, NR2F1, NRL, NYX, OAT, OFD1, OPA1, OPA3, OTX2, P3H2, PANK2, PAX2, PCDH15, PCYT1A, PDE6A, PDE6B, PDE6C, PDE6D, PDE6G, PDE6H, PDZD7, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHYH, PISD, PITPNM3, PLA2G5, PNPLA6, POC1B, POMGNT1, PRCD, PRDM13, PROM1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, PRPS1, RAB28, RAX2, RBP3, RBP4, RCBTB1, RD3, RDH11, RDH12, RDH5, REEP6, RGR, RGS9, RGS9BP, RHO, RIMS1, RLBP1, ROM1, RP1, RP1L1, RP2, RPE65, RPGR, RPGRIP1, RPGRIP1L, RS1, RTN4IP1, SAG, SAMD11, SCAPER, SCLT1, SDCCAG8, SEMA4A, SLC24A1, SLC25A46, SLC7A14, SNRNP200, SPATA7, SPP2, SRD5A3, TCTN1, TCTN2, TCTN3, TEAD1, TIMM8A, TIMP3, TMEM107, TMEM126A, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TOPORS, TRAF3IP1, TREX1, TRIM32, TRPM1, TSPAN12, TTC21B, TTC8, TTLL5, TTPA, TUB, TUBB4B, TULP1, USH1C, USH1G, USH2A, VCAN, VPS13B, WDPCP, WDR19, WFS1, YME1L1, ZNF408, ZNF423, ZNF513

**17. Nöro-  
Oftalmoloji Paneli  
Genleri;**

- ✓ ACO2, AFG3L2, ANTXR1, APTX, ATAD3A, AUH, C10ORF2, C12ORF65, C19ORF12, CHN1, CISD2, DNAJC19, DNMI1L, FDXR, FRMD7, GPR143, HESX1, KIF21A, MECR, MFN2, 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, MTPAP, NDUFS1, NR2F1, OPA1, OPA3, OTX2, PAX6, PHOX2A, POLG, PRPS1, ROBO3, RRM2B, RTN4IP1, SALL4, SETX, SLC25A4, SLC25A46, SLC38A8, SOX2, SPG7, TIMM8A, TK2, TMEM126A, TSFM, TUBB3, TYMP, UCHL1, WFS1, YME1L1, ZNHIT3

**18. Optik Atrofi  
Paneli Genleri;**

- ✓ ACO2, AFG3L2, ATAD3A, AUH, C12ORF65, C19ORF12, CISD2, DNAJC19, DNMI1L, FDXR, MECR, MFN2, 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, MTPAP, NDUFS1, NR2F1, OPA1, OPA3, POLG, PRPS1, RTN4IP1, SLC25A46, SLC52A2, SNX10, SPG7, TIMM8A, TMEM126A, TSFM, UCHL1, WFS1, YME1L1, ZNHIT3

## 19. Retinal Distrofi Paneli Genleri;

- ✓ ABCA4, ABCC6, ABHD12, ACO2, ADAM9, ADAMTS18, ADGRV1, ADIPOR1, AGBL5, AHII, AIPL1, ALMS1, ARHGEF18, ARL13B, ARL2BP, ARL3, ARL6, ARMC9, ARSG, ATF6, ATOH7, B9D1, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BEST1, C1QTNF5, C21ORF2, C2ORF71, C5ORF42, C8ORF37, CA4, CABP4, CACNA1F, CACNA2D4, CAPN5, CC2D2A, CDH23, CDH3, CDHR1, CEP104, CEP120, CEP164, CEP19, CEP250, CEP290, CEP41, CEP78, CERKL, CHM, CIB2, CISD2, CLN3, CLRN1, CNGA1, CNGA3, CNGB1, CNGB3, CNNM4, COL11A1, COL11A2, COL18A1, COL2A1, COL9A1, COL9A2, COL9A3, CPE, CRB1, CRX, CSPP1, CTC1, CTNNA1, CTNNB1, CWC27, CYP4V2, DFNB31, DHDDS, DHX38, DRAM2, DTHD1, EFEMP1, ELOVL4, EMC1, ESPN, EYS, FAM161A, FDXR, FLVCR1, FRMD7, FZD4, GNAT1, GNAT2, GNB3, GNPTG, GPR179, GRK1, GRM6, GUCA1A, GUCY2D, HARS, HGSNAT, HK1, HMX1, IDH3A, IDH3B, IFT140, IFT172, IFT27, IFT81, IMPDH1, IMPG1, IMPG2, INPP5E, INVS, IQCB1, JAG1, KCNJ13, KCNV2, KIAA0556, KIAA0586, KIAA0753, KIAA1549, KIF11, KIF7, KIZ, KLHL7, LCA5, LRAT, LRIT3, LRP2, LRP5, LZTFL1, MAK, MERTK, MFN2, MFRP, MFSD8, MKKS, MKS1, MMACHC, 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, MTPP, MVK, MYO7A, NDP, NEK2, NMNAT1, NPHP1, NPHP3, NPHP4, NR2E3, NR2F1, NRL, NYX, OAT, OFD1, OPA1, OPA3, OTX2, P3H2, PANK2, PAX2, PCDH15, PCYT1A, PDE6A, PDE6B, PDE6C, PDE6D, PDE6G, PDE6H, PDZD7, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHYH, PISD, PITPNM3, PLA2G5, PNPLA6, POC1B, POMGNT1, PRCD, PRDM13, PROM1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, PRPS1, RAB28, RAX2, RBP3, RBP4, RCBTB1, RD3, RDH11, RDH12, RDH5, REEP6, RGR, RGS9, RGS9BP, RHO, RIMS1, RLBP1, ROM1, RP1, RP1L1, RP2, RPE65, RPGR, RPGRIP1, RPGRIP1L, RS1, RTN4IP1, SAG, SAMD11, SCAPER, SCLT1, SDCCAG8, SEMA4A, SLC24A1, SLC25A46, SLC7A14, SNRNP200, SPATA7, SPP2, SRD5A3, TCTN1, TCTN2, TCTN3, TEAD1, TIMM8A, TIMP3, TMEM107, TMEM126A, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TOPORS, TRAF3IP1, TREX1, TRIM32, TRPM1, TSPAN12, TTC21B, TTC8, TTLL5, TTPA, TUB, TUBB4B, TULP1, USH1C, USH1G, USH2A, VCAN, VPS13B, WDPCP, WDR19, WFS1, YME1L1, ZNF408, ZNF423, ZNF513

**20. Retinitis  
Pigmentosa Paneli  
Genleri;**

- ✓ ABCA4, ABHD12, ADIPOR1, AGBL5, AH11, AIPL1, ARHGEF18, ARL2BP, ARL3, ARL6, BBS1, BBS2, BEST1, C1QTNF5, C21ORF2, C2ORF71, C8ORF37, CA4, CDHR1, CEP290, CERKL, CHM, CLN3, CLRN1, CNGA1, CNGB1, CRB1, CRX, CTNNA1, CWC27, CYP4V2, DHDDS, DHX38, EYS, FAM161A, FLVCR1, GNPTG, GUCY2D, HGSNAT, HK1, IDH3A, IDH3B, IFT140, IMPDH1, IMPG2, INPP5E, KIAA1549, KIZ, KLHL7, LCA5, LRAT, MAK, MERTK, MFRP, 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, MVK, NEK2, NMNAT1, NR2E3, NRL, OAT, OFD1, PDE6A, PDE6B, PDE6G, PEX1, PEX2, PEX7, PHYH, PITPNM3, PLA2G5, POMGNT1, PRCD, PROM1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, RBP3, RBP4, RCBTB1, RDH12, RDH5, REEP6, RGR, RHO, RIMS1, RLBP1, ROM1, RP1, RP2, RPE65, RPGR, RPGRIP1, RS1, SAG, SAMD11, SCAPER, SCLT1, SEMA4A, SLC7A14, SNRNP200, SPATA7, SPP2, TOPORS, TTC8, TTPA, TUB, TULP1, USH1C, USH2A, VPS13B, WDR19, ZNF408, ZNF513

**21. Senior-Loken  
Sendromu Paneli  
Genleri;**

- ✓ CEP290, INVS, IQCB1, NPHP1, NPHP3, NPHP4, SCLT1, SDCCAG8, TRAF3IP1

**22. Septo-Optik  
Displazi Paneli  
Genleri;**

- ✓ HESX1, OTX2, PAX6, SOX2





**23. Stickler  
Sendrom Paneli  
Genleri;**

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



**24. Usher  
Sendromu Paneli  
Genleri;**

- ✓ ABHD12, ADGRV1, ARSG, CDH23, CEP250, CEP78, CIB2, CLRN1, DFNB31, ESPN, HARS, MYO7A, PCDH15, PDZD7, PEX1, PEX6, PRPS1, TUBB4B, USH1C, USH1G, USH2A



**25. Vitreoretinopati  
Paneli Genleri;**

- ✓ ATOH7, BEST1, CAPN5, COL11A1, COL11A2, COL18A1, COL2A1, COL9A1, COL9A2, COL9A3, CTC1, CTNNB1, FZD4, KCNJ13, KIF11, LRP5, NDP, NR2E3, P3H2, RCBTB1, RS1, TSPAN12, VCAN, ZNF408