

## NEFROLOJİ POLİKLİNİĞİ

Böbrek fonksiyonlarında meydana gelen değişimler, böbrek yetmezliği, vücutta iyon yetersizliği, albüminin idrara çok geçmesi, kistik böbrek oluşumu, tuza duyarlı hipertansiyon oluşumu, iç organlarda kalsiyum oksalat birikimi, çocukluk retina ve böbrek hastalığı gibi böbrekle ilişkili hastalıkların genetik olarak belirlenmesi için yapılan çalışmaların olduğu panellerdir.

### 1. Alport Sendromu Paneli Genleri;

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

### 2. 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

### 3. Bartter Sendromu Paneli Genleri;

✓ AP2S1, BSND, CASR, CLCNKA, CLCNKB, GNA11, KCNJ1, MAGED2, SLC12A1, SLC12A3

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

- ✓ EYA1, SIX1, SIX5, TFAP2A

**5. Siliopati Paneli Genleri;**

- ✓ ACVR2B, AH11, ALMS1, ANKS6, ARL13B, ARL6, ARMC9, B9D1, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C21ORF2, C2CD3, C5ORF42, C8ORF37, CC2D2A, CENPF, CEP104, CEP120, CEP164, CEP19, CEP290, CEP41, CEP83, CPE, CRB2, CSPP1, DCDC2, DDX59, DHCR7, DYNC2H1, DYNC2LI1, EVC, EVC2, FAM58A, GLI2, GLI3, GLIS2, HYLS1, IFT122, IFT140, IFT172, IFT27, IFT43, IFT52, IFT80, IFT81, INPP5E, INVS, IQCB1, KIAA0556, KIAA0586, KIAA0753, KIF14, KIF7, LEFTY2, LZTFL1, MAPKBP1, MKKS, MKS1, NEK1, NEK8, NODAL, NPHP1, NPHP3, NPHP4, OFD1, PDE6D, PKD1, PKD2, PKHD1, PMM2, PNPLA6, POC1B, RPGRIP1L, SCAPER, SCLT1, SDCCAG8, TCTEX1D2, TCTN1, TCTN2, TCTN3, TMEM107, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TRAF3IP1, TRIM32, TTC21B, TTC8, USP9X, WDPCP, WDR19, WDR34, WDR35, WDR60, ZIC3, ZNF423

**6. Kistik Böbrek Hastalığı Paneli Genleri;**

- ✓ ANKS6, CEP164, CEP290, CEP83, COL4A1, CRB2, DCDC2, DNAJB11, DZIP1L, EYA1, GANAB, GLIS2, HNF1B, IFT172, INVS, IQCB1, JAG1, LRP5, MAPKBP1, NEK8, NOTCH2, NPHP1, NPHP3, NPHP4, OFD1, PAX2, PKD1, PKD2, PKHD1, RPGRIP1L, SDCCAG8, SEC61A1, SIX5, TMEM67, TSC1, TSC2, TTC21B, UMOD, VHL, WDR19, ZNF423

- 7. Diyabet İnsipidus Paneli Genleri;**
  - ✓ AQP2, AVP, AVPR2
- 8. Hemolitik Üremik Sendrom Paneli Genleri;**
  - ✓ ADAMTS13, C3, CD46, CFB, CFH, CFHR5, CFI, DGKE, THBD
- 9. Hipomagnezemi Paneli Genleri;**
  - ✓ BSND, CASR, CLCNKB, CLDN16, CLDN19, CNNM2, CNNM4, EGF, FAM111A, FXVD2, HNF1B, KCNA1, KCNJ10, MAGT1, NIPA2, PCBD1, SARS2, SLC12A3, TRPM6
- 10. Hipofosfatemik Raşitizm Paneli Genleri;**
  - ✓ ALPL, CLCN5, CYP27B1, CYP2R1, DMP1, ENPP1, FAH, FGF23, KL, PHEX, SLC34A1, SLC34A3, VDR
- 11. 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
- 12. Liddle Sendromu Paneli Genleri;**
  - ✓ SCNN1B, SCNN1G

**13. Meckel  
Sendromu Paneli  
Genleri;**

- ✓ B9D1, B9D2, CC2D2A, CEP290, KIF14, MKS1, NPHP3, RPGRIP1L, TCTN2, TMEM107, TMEM216, TMEM231, TMEM67

**14. Monogenik  
Obezite Paneli  
Genleri;**

- ✓ ADCY3, ALMS1, ARL6, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CEP19, CEP290, CPE, CUL4B, DYRK1B, GNAS, KSR2, LEP, LEPR, MAGEL2, MC3R, MC4R, MKKS, MKS1, NR0B2, NTRK2, PCSK1, PHF6, PHIP, POMC, PPARG, SDCCAG8, SH2B1, SIM1, TRIM32, TTC8, TUB, UCP3, VPS13B, WDPCP

**15. Nefrolityazis  
Paneli Genleri;**

- ✓ ADCY10, AGXT, ALPL, APRT, ATP6V0A4, ATP6V1B1, CA2, CASR, CLCN5, CLDN16, CLDN19, CYP24A1, FAM20A, GNA11, GPHN, GRHPR, HNF4A, HOGA1, HPRT1, KCNJ1, MOCOS, MOCS1, OCRL, SLC12A1, SLC22A12, SLC26A1, SLC2A9, SLC34A1, SLC34A3, SLC3A1, SLC4A1, SLC7A9, SLC9A3R1, VDR, XDH

**16. Nefronoftizis  
Paneli Genleri;**

- ✓ ANKS6, CEP164, CEP290, CEP83, DCDC2, GLIS2, IFT172, INVS, IQCB1, MAPKBP1, NEK8, NPHP1, NPHP3, NPHP4, RPGRIP1L, SDCCAG8, TMEM67, TTC21B, WDR19, ZNF423

**17. Nefrotik  
Sendrom Paneli  
Genleri;**

- ✓ ACTN4, ADCK4, ANLN, APOL1, ARHGAP24, ARHGDI, CD2AP, COL4A3, COL4A4, COL4A5, COQ2, CRB2, DGKE, DLC1, EMP2, FAN1, FAT1, FN1, INF2, ITGA3, KANK1, KANK2, KANK4, LAMB2, LMX1B, MAFB, MAGI2, 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, MYO1E, NPHS1, NPHS2, NUP107, NUP93, OSGEP, PLCE1, PTPRO, SCARB2, SGPL1, SMARCAL1, TRPC6, TTC21B, WDR73, WT1, XPO5

**18. Polikistik  
Böbrek Hastalığı  
Paneli Genleri;**

- ✓ DNAJB11, DZIP1L, GANAB, HNF1B, JAG1, LRP5, NOTCH2, PKD1, PKD2, PKHD1, PRKCSH, SEC63

**19. Primer Siliyer  
Diskinezi Paneli  
Genleri;**

- ✓ ARMC4, C11ORF70, C21ORF59, CCDC103, CCDC114, CCDC39, CCDC40, CCDC65, CCNO, CENPF, CFTR, DNAAF1, DNAAF2, DNAAF3, DNAAF5, DNAH1, DNAH11, DNAH5, DNAH9, DNAI1, DNAI2, DNAL1, DRC1, DYX1C1, GAS2L2, GAS8, HYDIN, INVS, LRRC6, MCIDAS, NME8, OFD1, PIH1D3, RPGR, RSPH1, RSPH3, RSPH4A, RSPH9, SPAG1, STK36, ZMYND10

- 20. Primer Hiperoksalüri Paneli Genleri;**
  - ✓ AGXT, GRHPR, HOGA1
  
- 21. Psödohipoaldosteronizm Paneli Genleri;**
  - ✓ CUL3, HSD11B2, KCNJ5, KLHL3, NR3C2, SCNN1A, SCNN1B, SCNN1G, WNK1, WNK4
  
- 22. Böbrek Malformasyonu Paneli Genleri;**
  - ✓ ACE, ACTG2, AGT, BMP4, CDC5L, CHD1L, DSTYK, EYA1, FAM58A, FANCB, FOXC2, FREM1, FREM2, GATA3, GREB1L, HNF1B, KYNU, PAX2, PBX1, PUF60, REN, RET, ROBO2, SALL1, SIX1, SIX5, WT1
  
- 23. Renal Tübüler Asidoz Paneli Genleri;**
  - ✓ ATP6V0A4, ATP6V1B1, CA2, SLC4A1, SLC4A4
  
- 24. Senior-Loken Sendromu Paneli**
  - ✓ CEP290, INVS, IQCB1, NPHP1, NPHP3, NPHP4, SCLT1, SDCCAG8, TRAF3IP1