

## ENDOKRİNOLOJİ VE GENETİK POLİKLİNİĞİ

Belirsiz dış genital özellikler, şeker ve obezite ilişkisi, karbonhidrat metabolizması bozuklukları, tiroid ve paratiroid bezlerinin çalışma şekillerine bağlı meydana gelen cinsel ve vücutl gelişim bozuklukları, insüline bağımlı olmayan diyabet çeşitleri, enerji metabolizmasında meydana gelen değişimler gibi endokrinolojiyi ilgilendiren ve tedaviyi kolaylaştırıp yenidoğan ünitelerinde ve prenatal tanıda erken teşhis oluşturabilmek için yapılan çalışmaların olduğu panellerdir.

### 1. Ambigus Genitale / Cinsel Gelişim Bozuklukları Paneli Genleri;

- ✓ AMH, AMHR2, ANOS1, AR, ARX , ATRX, BCOR, CDK9, CDKN1C, CEP41, CHD7, CREBBP, CYB5A, CYP11A1, CYP11B1 , CYP17A1, CYP19A1, CYP21A2, DHCR7, DHH, DYNC2H1, ERCC3, FEZF1, FGF8, FGFR1, FIG4, FRAS1, GATA4 , GNRH1, GNRHR, HSD17B3, HSD3B2, IL17RD, IRF6, KISS1, KISS1R, LHB , LHCGR, MAMLD1, MAP3K1, MKRN3, MKS1, NR0B1, NR5A1, POR, PROK2, PROKR2, RSPO1, SAMD9, SGPL1, SOX10, SOX9, SRD5A2, SRY, STAR, TAC3, TACR3, TOE1, TSPYL1, WDR11, WT1, ZFPM2

### 2. Kapsamlı Monogenik Diyabet Paneli Genleri;

- ✓ ABCC8, APPL1, BLK, CEL, EIF2AK3, FOXP3, GATA6, GCK, GLIS3, GLUD1, HADH, HNF1A, HNF1B, HNF4A, INS, INSR, KCNJ11, KLF11, 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, NEUROD1, NEUROG3, PAX4, PDX1, PPARG, PTF1A, RFX6, SLC16A1, SLC2A2, UCP2, WFS1, ZFP57

- 3. Konjenital Adrenal Hiperplazi Paneli Genleri;**
  - ✓ ARMC5, CYP11A1, CYP11B1 , CYP11B, CYP17A1, CYP21A2 , HSD3B2, PDE11A, PDE8B, POR, PRKAR1A, STAR
- 4. Glukokortikoid Eksiklik Paneli Genleri;**
  - ✓ AAAS, MC2R, MRAP, NNT, NR3C1, POMC, STAR
- 5. Hiperlipidemi Paneli Genleri;**
  - ✓ ABCA1, ABCG5, ABCG8, ALMS1 , APOA1, APOA5, APOB, APOC2, APOC3, APOE, CREB3L3, GPIHBP1, LDLR, LDLRAP1, LIPA, LMF1, LPL, PCSK9
- 6. Hiperparatiroidizm Paneli Genleri;**
  - ✓ AIRE, AP2S1, CASR, CDC73, CDKN1A, CDKN1B, CDKN2B, CDKN2C, GCM2, GNA11, MEN1, PTH, RET, TRPV6
- 7. Hipoglisemi, Hiperinsülinizm ve Keton Metabolizması Paneli Genleri;**
  - ✓ ABCC8, ACAT1, ACSF3, AGL, ALDOA, ALDOB, ENO3, EPM2A, FBP1, G6PC, GAA, GBE1, GCK, GLUD1 , GYG1, GYS1, GYS2, HADH, HMGCL, HMGCS2, HNF1A, HNF4A, INSR, KCNJ11, LAMP2, LDHA, MPV17, NHLRC1, OXCT1, PC, PCK1, PDX1, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PRKAG2, PRKAG3, PTF1A, PYGL, PYGM, RBCK1, SLC16A1, SLC2A2, SLC37A4, UCP6

**8. Hipomagnezemi  
Paneli Genleri;**

- ✓ BSND, CASR, CLCNKB , CLDN16, CLDN19, CNNM2, CNNM4, EGF, FAM111A, FXVD2, HNF1B, KCNA1, KCNJ10, MAGT1, NIPA2, PCBD1, SARS2, SLC12A3, TRPM6

**9. Hipotiroidizm ve  
Tiroid Hormon Direnci  
Paneli Genleri;**

- ✓ DUOX2 , DUOXA2, FOXE1, GNAS, HESX1, IGSF1, NKX2-1, NKX2-5, PAX8, POU1F1, PROP1, SECISBP2, SLC16A2, SLC26A4, SLC5A5, TBL1X, TG, THRA, THRB, TPO, TSHB, TSHR

**10. Kallmann  
Sendromu Paneli  
Genleri;**

- ✓ ANOS1 , CDK9, CHD7, FEZF1, FGF8, FGFR1, GNRH1, GNRHR, HS6ST1 , KISS1, KISS1R, PROK2, PROKR2, SOX10, TAC3, TACR3, WDR11

**11. MODY Paneli  
Genleri;**

- ✓ ABCC8, APPL1, BLK, CEL , GCK, HNF1A, HNF1B, HNF4A, INS, KCNJ11, KLF11, 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, NEUROD1, PAX4, PDX1, RFX6



**12. 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



**13. Prematür Ovaryen  
Yetmezlik Paneli  
Genleri;**

- ✓ BMP15, CYP17A1, CYP19A1, FOXL2, FSHR, GALT, GNAS, LHCGR, LMNA, NOBOX, NR5A1, POLG, POR, STAG3 , STAR, WT1