

MİTOKONDRIAL HASTALIKLAR VE GENETİK POLİKLİNİĞİ

Mitokondrial DNA içeriğinin azalması, glisin yıkımında meydana gelen bozukluklar, periferik sinir sistemi bozuklukları, bazı aminoasitlerin metabolizmalarında meydana gelen bozukluklar, böbrek taşı oluşumu gibi mitokondrial yapıda meydana gelen ve vücudu etkileyen bozuklukları belirleyip bu sonuçları prenatal tanıda kullanmak için yapılan çalışmaların olduğu panellerdir.

1. Mitokondriyal DNA Deplezyon Sendromu Paneli Genleri;

✓ AGK, APTX, AUH, C10ORF2, C12ORF65, DGUOK, FBXL4, MFN2, MPV17, 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, MTTV, MTTW, MTTY, NDUFS1, OPA1, OPA3, POLG, POLG2, RRM2B, SLC25A3, SLC25A4, SPG7, SUCLA2, SUCLG1, TIMM8A, TK2, TMEM126A, TYMP, WFS1

2. 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

**3. 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

**4. Nonketotik
Hiperglisemi / Glisin
Ensefalopati Paneli
Genleri;**

- ✓ AMT, BOLA3, GCSH, GLDC, GLRX5, LIAS, LIPT1, NFU1, SLC6A9

**5. Organik Asidemi /
Asidüri ve Kobalamin
Eksikliği Paneli
Genleri;**

- ✓ ABCD4, ACADSB, ACAT1, ACSF3, ADK, AHCY, AMN, BCKDHA, BCKDHB, BCS1L, CBS, CD320, CLPB, CTH, CUBN, D2HGDH, DBT, DLD, ETFA, ETFB, ETFDH, FLAD1, GCDH, GIF, GNMT, HCFC1, HIBCH, HMGCL, IDH2, IVD, L2HGDH, LMBRD1, MCCC1, MCCC2, MCEE, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MTHFR, MTR, MTRR, MUT, PCCA, PCCB, PEPD, SERAC1, SLC25A1, SUCLA2, SUCLG1, SUGCT, TCN2, UMPS

**6. Periyodik Felç Paneli
Genleri;**

- ✓ CACNA1S, CLCN1, KCNJ2, SCN4A

**7. Peroksizomal
Bozukluklar Paneli
Genleri;**

- ✓ ABCD1, ABCD3, ACOX1, AGPS, AGXT, AMACR, DYM, EBP, GNPAT, HSD17B4, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHYH, SUGCT, TRIM37

**8. Porfiria Paneli
Genleri;**

- ✓ ALAD, ALAS2, CPOX, FECH, HFE, HMBS, PPOX, UROD, UROS

**9. Pürin ve Pirimidin
Metabolizması
Bozuklukları Paneli
Genleri;**

- ✓ ADA, ADSL, AMPD1, APRT, ATIC, DHODH, DPYD, DPYS, GPHN, HPRT1, MOCOS, MOCS1, NT5C3A, PNP, PRPS1, REN, TPMT, UMOD, UMPS, UPB1, XDH

**10. Tirozinemi Paneli
Genleri;**

- ✓ AGK, APTX, AUH, C10ORF2, C12ORF65, DGUOK, FBXL4, MFN2, MPV17, 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, MTTV, MTTW, MTTY, NDUFS1, OPA1, OPA3, POLG, POLG2, RRM2B, SLC25A3, SLC25A4, SPG7, SUCLA2, SUCLG1, TIMM8A, TK2, TMEM126A, TYMP, WFS1