

## METABOLİK HASTALIKLAR VE GENETİK POLİKLİNİĞİ

Yağ, karbonhidrat metabolizması bozuklukları, karaciğer yağlanması, demir metabolizması bozukluğu, idrarda artmış homosistin, lizozom depo hastalıkları metabolizması, kalsifikasyon sonucu oluşan genetik rahatsızlıklar gibi vücutta meydana gelen metabolik rahatsızlıkları incelemek ve anlamlandırmak için yapılan çalışmaların olduğu panellerdir.

### 1. Aicardi-Goutières Sendromu Paneli Genleri;

✓ ADAR, IFIH1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, TREX1

### 2. Koenzim q10 Eksiklik Paneli Genleri;

✓ ADCK3, ANO10, APTX, COQ2, COQ4, COQ5, COQ6, COQ7, COQ9, ETFA, ETFB, ETFDH, PDSS1, PDSS2, SLC25A26

### 3. Konjenital ve Ailesel Lipodistrofi Paneli Genleri;

✓ AGPAT2, AKT2, BSCL2, CAV1, CIDEC, LIPE, LMNA, PLIN1, PPARG, PTRF, TBC1D4, ZMPSTE24

**4. Konjenital  
Glikozilasyon  
Bozuklukları Paneli  
Genleri;**

- ✓ ALG1, ALG11, ALG12, ALG13, ALG2, ALG3, ALG6, ALG8, ALG9, ATP6V0A2, B3GLCT, B4GALT1, COG1, COG4, COG5, COG6, COG7, COG8, DDOST, DHDDS, DOLK, DPAGT1, DPM1, DPM2, DPM3, FUT8, GMPPA, GNE, MAGT1, MAN1B1, MGAT2, MOGS, MPDU1, MPI, NGLY1, PGM1, PMM2, RFT1, SEC23B, SLC35A1, SLC35A2, SLC35C1, SRD5A3, SSR4, STT3A, STT3B, TMEM165, TUSC3

**5. Konjenital Mono ve  
Disakkarid  
Bozuklukları Paneli  
Genleri;**

- ✓ ALDOB, GALE, GALK1, GALT, LCT, SI, SLC2A1, SLC2A2, SLC5A1

**6. Kreatin  
Metabolizması  
Eksikliği Paneli  
Genleri;**

- ✓ GAMT, GATM, SLC6A8

✓ ABCC8, ABCD1, ABCD3, ABCD4, ACAD8, ACAD9, ACADL, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACOX1, ACSF3, ACY1, ADA, ADAMTSL2, ADAR, ADCK3, ADK, ADSL, AGA, AGK, AGL, AGPAT2, AGPS, AGXT, AHCY, AKT2, ALAD, ALAS2, ALDH5A1, ALDH7A1, ALDOA, ALDOB, ALG1, ALG11, ALG12, ALG13, ALG2, ALG3, ALG6, ALG8, ALG9, AMACR, AMN, AMPD1, AMT, ANO10, ANO5, ANTXR2, APRT, APTX, ARG1, ARSA, ARSB, ASAH1, ASL, ASPA, ASS1, ATIC, ATP13A2, ATP6V0A2, ATP7B, AUH, B3GLCT, B4GALT1, BCKDHA, BCKDHB, BCS1L, BOLA3, BSCL2, BSND, BTD, C10ORF2, C12ORF65, CA5A, CACNA1S, CASR, CAV1, CAV3, CBS, CD320, CIDEC, CLCN1, CLCNKB, CLDN16, CLDN19, CLN3, CLN5, CLN6, CLN8, CLPB, CNNM2, CNNM4, COG1, COG4, COG5, COG6, COG7, COG8, COL11A2, COL2A1, COQ2, COQ4, COQ5, COQ6, COQ7, COQ9, CPOX, CPS1, CPT1A, CPT2, CTH, CTNS, CTSA, CTSC, CTSD, CTSK, CUBN, D2HGDH, DBT, DDOST, DGUOK, DHCR7, DHDDS, DHODH, DLD, DNAJC12, DNMI1, DOLK, DPAGT1, DPM1, DPM2, DPM3, DPYD, DPYS, DYM, DYSF, EBP, ECHS1, EGF, ENO3, EPM2A, ETFA, ETFB, ETFDH, FAH, FAM111A, FBP1, FBXL4, FECH, FH, FKRP, FKTN, FLAD1, FLNA, FLNB, FMO3, FOLR1, FOXRED1, FUCA1, FUT8, FXYD2, G6PC, GAA, GALC, GALE, GALK1, GALNS, GALT, GAMT, GATM, GBA, GBE1, GCDH, GCH1, GCK, GCSH, GFM1, GIF, GLA, GLB1, GLDC, GLRX5, GLUD1, GLUL, GM2A, GMPPA, GNE, GNMT, GNPAT, GNPTAB, GNPTG, GNS, GPC3, GPHN, GUSB, GYG1, GYS1, GYS2, HADH, HADHA, HADHB, HAMP, HCFC1, HEXA, HEXB, HFE, HFE2, HGD, HGSNAT, HIBCH, HLCS, HMBS, HMGCL, HMGCS2, HNF1A, HNF1B, HNF4A, HPD, HPRT1, HRAS, HSD17B10, HSD17B4, HYAL1, IDH2, IDS, IDUA, IFIH1, INSR, ISCU, IVD, KCNA1, KCNJ10, KCNJ11, KCNJ2, L2HGDH, LAMA2, LAMP2, LCT, LDB3, LDHA, LIAS, LIPA, LIPE, LIPT1, LMBRD1, LMNA, LPIN1, MAGT1, MAN1B1, MAN2B1, MANBA, MCCC1, MCCC2, MCEE, MCOLN1, MFN2, MFSD8, MGAT2, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MOCOS, MOCS1, MOCS2, MOGS, MPDU1, MPI, 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, MT-TV, MT-TW, MT-TY, MTHFR, MTR, MTRR, MUT, MYH3, MYOT, NAGA, NAGLU, NAGS, NBAS, NDUFAF2, NDUFS1, NEU1, NFU1, NGLY1, NHLRC1, NIPA2, NPC1, NPC2, NT5C3A, OAT, OPA1, OPA3, OTC, OXCT1, PAH, PC, PCBD1, PCCA, PCCB, PCK1, PDHA1, PDHB, PDHX, PDSS1, PDSS2, PDX1, PEPD, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PHYH, PLIN1, PMM2, PNP, PNPLA2, POLG, POLG2, PPARG, PPOX, PPT1, PRKAG2, PRKAG3, PRODH, PRPS1, PSAP, PTF1A, PTRF, PTS, PYGL, PYGM, QDPR, RAI1, RBCK1, REN, RFT1, RNASEH2A, RNASEH2B, RNASEH2C, RRM2B, RYR1, SAMHD1, SARS2, SCN4A, SEC23B, SERAC1, SERPINA1, SGSH, SI, SLC12A3, SLC16A1, SLC17A5, SLC22A5, SLC25A1, SLC25A10, SLC25A13, SLC25A15, SLC25A20, SLC25A26, SLC25A3, SLC25A4, SLC2A1, SLC2A2, SLC30A1, SLC35A1, SLC35A2, SLC35C1, SLC37A4, SLC39A4, SLC3A1, SLC40A1, SLC46A1, SLC5A1, SLC6A19, SLC6A8, SLC6A9, SLC7A7, SLC7A9, SMPD1, SPG7, SRD5A3, SSR4, STT3A, STT3B, SUCLA2, SUCLG1, SUGCT, SUMF1, SUOX, TALDO1, TANGO2, TAT, TAZ, TBC1D4, TCF4, TCN2, TFR2, TIMM8A, TK2, TMEM126A, TMEM165, TMEM70, TPMT, TPP1, TREX1, TRIM37, TRPM6, TUSC3, TYMP, UCP2, UMOD, UMPS, UPB1, UROD, UROS, WFS1, XDH, ZMPSTE24

## 7. Kapsamlı Metabolizma Paneli Genleri;

**8. Sistinüri Paneli Genleri;**

✓ SLC3A1, SLC7A9

**9. Yağ Asidi Oksidasyon Sendromu Paneli Genleri;**

✓ ACAD8 ,ACAD9, ACADL, ACADM, ACADS, ACADSB, ACADVL, ALDH5A1, CPT1A, CPT2, ECHS1, ETFA, ETFB, ETFDH, GLUD1, HADH, HADHA, HADHB, HMGCL, HMGCS2, HSD17B10, LPIN1, PPARG, SLC22A5, SLC25A20, TAZ

**10. Glikojen Depolama Bozukluğu Paneli Genleri;**

✓ AGL ,ALDOA, ENO3, EPM2A, FBP1, G6PC, GAA, GBE1, GYG1, GYS1, GYS2, LAMP2, LDHA, NHLRC1, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PRKAG2, PRKAG3, PYGL, PYGM, RBCK1, SLC2A2, SLC37A4

**11. Kalıtsal Hemokromatoz Paneli Genleri;**

✓ HAMP, HFE, HFE2, SLC40A1, TFR2

**12. Homosistinüri Çekirdek Paneli Genleri;**

✓ CBS, MTHFR, MTR, MTRR

**13. Hiperammonemi ve Üre Döngüsü Bozukluk Paneli Genleri;**

- ✓ ACADM, ACADS, ACADVL, ARG1, ASL, ASS1, BCKDHA, BCKDHB, CA5A, CPS1, CPT1A, CPT2, DBT, DLD, ETFB, ETFDH, GLUD1, GLUL, HADHA, HADHB, HCFC1, HLCS, HMGCL, HMGCS2, IVD, MCCC1, MCCC2, MMAA, MMAB, MMACHC, MMADHC, MUT, NAGS, NBAS, OAT, OTC, PC, PCCA, PCCB, SLC22A5, SLC25A13, SLC25A15, SLC25A20, SLC7A7, SUCLA2, SUCLG1, TMEM70, UMPS

**14. Hiperfenilalaninemi Paneli Genleri;**

- ✓ DNAJC12, GCH1, PAH, PCBD1, PTS, QDPR

**15. 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, UCP2

**16. Hipomagnezemi Paneli Genleri;**

- ✓ BSND, CASR, CLCNKB, CLDN16, CLDN19, CNNM2, CNNM4, EGF, FAM111A, FXD2, HNF1B, KCNA1, KCNJ10, MAGT1, NIPA2, PCBD1, SARS2, SLC12A3, TRPM6

**17. Lizozomal  
Bozukluklar ve  
Mukopolisakkaridoz  
Paneli Genleri;**

- ✓ ABCC8, ACY1, ADAMTSL2, ADSL, AGA, ALDH5A1, ALDH7A1, AMT, ANTXR2, ARG1, ARSA, ARSB, ASAH1, ASPA, ATP13A2, BTBD9, CLN3, CLN5, CLN6, CLN8, COL11A2, COL2A1, CTNS, CTSA, CTSC, CTSD, CTSK, DHCR7, DPYD, DYM, ETFA, ETFB, ETFDH, FH, FOLR1, FUCA1, GAA, GALC, GALNS, GAMT, GBA, GCDH, GLA, GLB1, GLDC, GM2A, GNE, GNPTAB, GNPTG, GNS, GPC3, GUSB, HEXA, HEXB, HGSNAT, HRAS, HYAL1, IDS, IDUA, L2HGDH, LAMA2, LAMP2, LDB3, LIPA, MAN1B1, MAN2B1, MANBA, MCOLN1, MFSD8, MOCS1, MOCS2, MYOT, NAGA, NAGLU, NEU1, NPC1, NPC2, PEX1, PEX10, PEX12, PEX13, PEX16, PEX26, PEX3, PEX5, PEX6, PGK1, PHYH, PPT1, PRODH, PSAP, QDPR, RAI1, SGSH, SLC17A5, SLC25A15, SLC46A1, SMPD1, SUMF1, SUOX, TCF4, TPP1

**18. Metabolik  
Karaciğer Yetmezliği  
Paneli Genleri;**

- ✓ ALDOB, ATP7B, FAH, GALE, GALK1, GALT, LIPA, MPI, NPC1, NPC2, PHKA2, PHKB, PHKG2, PYGL, SERPINA1, SMPD1

**19. Metabolik Miyopati  
ve Rabdomiyoliz Paneli  
Genleri;**

- ✓ ACAD9, ACADL, ACADM, ACADVL, ADCK3, AGL, AHCY, ALDOA, AMPD1, ANO5, C10ORF2, CAV3, COQ2, CPT2, DYSF, ENO3, ETFA, ETFB, ETFDH, FKRP, FKTN, FLAD1, GAA, GBE1, GYG1, GYS1, HADHA, HADHB, ISCU, LDHA, LPIN1, 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, MYH3, OPA1, OPA3, PFKM, PGAM2, PGK1, PGM1, PHKA1, POLG, POLG2, PYGM, RBCK1, RRM2B, RYR1, SCN4A, SLC22A5, SLC25A20, SUCLA2, SUCLG1, TANGO2, TK2, TYMP