

## NÖROLOJİ VE GENETİK POLİKLİNİĞİ

Vücut kaslarında meydana gelen güçsüzlük, miyopati, kortikal malformasyon, epilepsi, Parkinson, SMA(Spinal Muskuler Atrofi), denge güçlüğü, bunama, erken beyin gelişimi, migren gibi nörolojik durumların genetikle bağlantısını ortaya çıkarmak, sonraki nesillerin nasıl olacağına dair fikir yürütmek ve analizini yapıp multidisipliner çalışmaya katkı sağlamak amacıyla yapılan çalışmaların olduğu panellerdir.

### 1. Amyotrofik Lateral Sklerozis Paneli Genleri;

- ✓ ALS2, ANG, ATL1, BSCL2, CHCHD10, CHMP2B, DCTN1, FIG4, FUS, GBE1, GRN, HEXA, HNRNPA1, HSPD1, KIAA0196, KIF5A, MATR3, OPTN, PFN1, PRF1, REEP1, SETX, SLC52A2, SLC52A3, SOD1, SPAST, SPG11, SPG20, SQSTM1, TARDBP, TIA1, TUBA4A, UBQLN2, VAPB, VCP

## 2. Ataksi Paneli Genleri;

- ✓ ABCB7, ABHD12, ACO2, ADCK3, ADPRHL2, AFG3L2, AGTPBP1, AHII1, ALDH5A1, ANO10, APTX, ARL13B, ARL6, ATCAY, ATM, ATP1A3, ATP2B3, ATP8A2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BEAN1, C10ORF2, C12ORF4, C5ORF42, CA8, CACNA1A, CACNA1G, CACNB4, CAMTA1, CAPN1, CASK, CC2D2A, CCDC88C, CEP290, CEP41, CLCN2, CLN5, CLPP, COA7, COASY, COX20, CP, CSTB, CWF19L1, CYP27A1, CYP2U1, DHPS, DNAJC19, DNMT1, DOCK3, EBF3, EEF2, ELOVL4, ELOVL5, FA2H, FBXL4, FDXR, FGF14, FLVCR1, FMR1, FXN, GBA2, GFAP, GOSR2, GRID2, GRM1, GSS, HARS2, HIBCH, INPP5E, IRF2BPL, ITM2B, ITPR1, KCNA1, KCNC3, KCND3, KCNJ10, KIF1C, KIF7, LAMA1, LARS2, LMNB1, LRPPRC, MARS2, MECR, MKKS, MKS1, MME, MRE11A, MSTO1, 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, MTFMT, MTPAP, MTPP, NDUFAF6, NDUFS2, NDUFS4, NDUFS7, NDUFS8, NDUFV1, NKX6-2, NOL3, NPHP1, NUBPL, OFD1, OPA1, OPHN1, PAX6, PDYN, PEX7, PHYH, PNKD, PNKP, PNPLA6, POLG, PRKCG, PRRT2, PUM1, RNF216, RPGRIP1L, RUBCN, SACS, SCYL1, SERAC1, SETX, SIL1, SLC1A3, SLC20A2, SLC25A46, SLC2A1, SLC52A2, SLC9A1, SLC9A6, SNX14, SPG7, SPTBN2, STUB1, SYNE1, SYT14, TCTN1, TCTN2, TCTN3, TDP1, TGM6, TMEM138, TMEM216, TMEM231, TMEM237, TMEM240, TMEM67, TPP1, TRIM32, TTBK2, TTC19, TTC8, TTPA, TUBB4A, UBA5, UBTF, UCHL1, VAMP1, VLDLR, WDPCP, WDR81, WFS1, WWOX, ZFYVE26, ZNF423

**3. Otizm  
Spektrum  
Bozukluğu Paneli  
Genleri;**

- ✓ ADNP, BCL11A, C12ORF4, CACNA1C, CC2D1A, CNOT3, CNTN6, COL4A3BP, CSNK2A1, CTNND2, DHCR7, EHMT1, EN2, FBXO11, FOXP1, GAMT, KMT2E, KMT5B, MBOAT7, MECP2, 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, NBEA, NFIB, NLGN3, NLGN4X, NSD1, POGZ, PTCHD1, PTEN, RPL10, SHANK3, TBR1, TCF20, TRIP12, TSC1, TSC2, VAMP2, WASF1, ZSWIM6

- ✓ ABAT, ABCA2, ABCD1, ACTL6B, ADAM22, ADAR, ADPRHL2, ADSL, AFG3L2, AGA, AIFM1, AIMP1, ALDH3A2, ALDH5A1, ALDH7A1, ALG13, ALKBH8, AMACR, AMT, ANKRD11, AP2M1, AP3B2, AP4B1, AP4E1, AP4M1, AP4S1, APOPT1, ARG1, ARHGEF9, ARID1B, ARSA, ARX, ASAH1, ASNS, ASPA, ASXL3, ATP13A2, ATP1A3, ATP6V1A, ATRX, BRAT1, BTBD, C12ORF57, CACNA1A, CACNA1B, CACNA1D, CACNA1E, CACNA1H, CACNB4, CASK, CASR, CC2D1A, CDK9, CDKL5, CERS1, CHD2, CHRNA2, CHRNA4, CHRNB2, CLCN2, CLCN4, CLN3, CLN5, CLN6, CLN8, CLTC, CNKSR2, CNPY3, CNTNAP2, COA7, COL4A1, COL4A3BP, COX15, COX6B1, CPT2, CSF1R, CSNK2B, CSTB, CTC1, CTSD, CTSF, CUL4B, CYFIP2, CYP27A1, D2HGDH, DARS, DARS2, DCX, DDC, DDX3X, DEGS1, DENND5A, DEPDC5, DHFR, DHPS, DNAJC5, DNM1, DNM1L, DOCK7, DPYD, DPYS, DYRK1A, EARS2, ECHS1, ECM1, EEF1A2, EFHC1, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF3F, EPM2A, EPRS, ETFA, ETFB, ETFDH, ETHE1, FA2H, FAM126A, FAR1, FARS2, FDF1T1, FDX1L, FGF12, FH, FLNA, FOLR1, FOXG1, FOXRED1, FRRS1L, FUT8, GABBR2, GABRA1, GABRB2, GABRB3, GABRG2, GALC, GAMT, GCDH, GCH1, GFAP, GFM1, GFM2, GJC2, GLB1, GLDC, GLRB, GNAO1, GNB1, GNE, GOLGA2, GOSR2, GPAA1, GPHN, GRIA3, GRIA4, GRIK2, GRIN1, GRIN2A, GRIN2B, GRN, GTPBP3, HACE1, HCN1, HCN2, HECW2, HEPACAM, HIBCH, HNRNPU, HSD17B10, HSPD1, HTRA1, HTT, IBA57, ICK, IQSEC2, IRF2BPL, KCNA1, KCNA2, KCNB1, KCNC1, KCNH1, KCNMA1, KCNQ2, KCNQ3, KCNQ5, KCNT1, KCNT2, KCTD3, KCTD7, KDM5C, KIAA2022, KIF1A, KMT2E, L2HGDH, LGII, LMNB1, LRPPRC, LYRM7, MACF1, MAGI2, MARS2, MBD5, MBOAT7, MDH2, MECP2, MED12, MED17, MEF2C, MFSD8, MIPEP, MLC1, MOCS1, MRPL44, 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, MTFMT, MTHFR, MTOR, NACC1, NBEA, NDST1, NDUFAF3, NDUFAF5, NDUFAF6, NDUFS2, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NECAP1, NEU1, NEUROD2, NFU1, NHLRC1, NKX6-2, NOTCH3, NPRL3, NRXN1, NT5C2, NUBPL, NUS1, OFD1, OPHN1, P4HTM, PARS2, PCDH19, PGK1, PHACTR1, PHF6, PIGA, PIGB, PIGC, PIGG, PIGN, PIGO, PIGP, PIGQ, PIGS, PIGT, PIGV, PIGW, PITRM1, PLAA, PLCB1, PLP1, PNKP, PNPO, POLG, POLR3A, POLR3B, PPP2CA, PPP3CA, PPT1, PRICKLE1, PRIMA1, PRODH, PROSC, PRRT2, PSAP, PTPN23, PTS, PUM1, PURA, PYCR2, QARS, QDPR, RAB39B, RALA, RARS, RELN, RMND1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF216, ROGDI, RORB, SAMHD1, SCARB2, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SCN9A, SCO1, SDHAF1, SERAC1, SERPIN1, SIK1, SLC12A5, SLC13A5, SLC19A3, SLC1A4, SLC25A1, SLC25A15, SLC25A22, SLC25A42, SLC2A1, SLC35A2, SLC39A8, SLC46A1, SLC6A1, SLC6A8, SLC9A6, SMARCA2, SMC1A, SMS, SNAP25, SNORD118, SOX10, SPATA5, SPTAN1, SPTBN4, ST3GAL3, ST3GAL5, STX1B, STXBP1, SUMF1, SUOX, SYN1, SYNGAP1, SYNJ1, SZT2, TAF1, TBC1D20, TBC1D24, TBCD, TBCE, TBCK, TBL1XR1, TCF4, TPK1, TPP1, TRAK1, TREX1, TRIM8, TSC1, TSC2, TTC19, TUBB4A, UBA5, UBE2A, UBE3A, UNC80, VAMP2, VARS, VPS13A, WARS2, WASF1, WDR26, WDR45, WWOX, YY1, ZEB2, ZFYVE26

**4. Avrupa ve  
Orta Doğu  
İçin; Pediatrik  
Epilepsi Paneli  
Genleri;**

**5. Serebral  
Kavernöz  
Malformasyon  
Paneli Genleri;**

- ✓ CCM2, KRIT1, PDCD10, RASA1

**6. Charcot-Marie-  
Tooth Nöropati  
Paneli Genleri;**

- ✓ AARS, AGTPBP1, AIFM1, AMACR, ARHGEF10, ATAD3A, ATL1, ATL3, ATP1A1, ATP7A, BAG3, BICD2, BSCL2, C12ORF65, CCT5, CHCHD10, COA7, COX10, COX6A1, CTDP1, DCAF8, DCTN1, DHTKD1, DNMT2, DNMT1, DST, DYNC1H1, EGR2, FAM134B, FBLN5, FGD4, FIG4, FXN, GAN, GARS, GDAP1, GJB1, GNB4, GNE, HADHB, HARS, HINT1, HK1, HSPB1, HSPB8, IGHMBP2, IKBKAP, INF2, KARS, KIF1A, KIF1B, KIF5A, LDB3, LITAF, LMNA, LRSAM1, MARS, MCM3AP, MED25, MFN2, MME, MORC2, MPZ, MTMR2, MYOT, NDRG1, NEFH, NEFL, NGF, NTRK1, PDK3, PLEKHG5, PMP22, POLG, PRDM12, PRPS1, PRX, RAB7A, REEP1, SACS, SBF1, SBF2, SCN11A, SCN9A, SCYL1, SEPT9, SETX, SH3TC2, SLC12A6, SLC25A46, SMAD3, SPG11, SPTBN4, SPTLC1, SPTLC2, SURF1, TFG, TRIM2, TRPV4, TTR, TYMP, VCP, WNK1, YARS, ZFYVE26

**7. Koenzim q10  
Eksiklik Paneli  
Genleri;**

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

**8. Kollajen Tip VI  
ile İlgili  
Bozukluklar  
Paneli Genleri;**

✓ COL12A1, COL4A1, COL4A2, COL6A1, COL6A2, COL6A3

**9. Kapsamlı Kas  
Distrofisi /  
Miyopati Paneli  
Genleri;**

✓ ACTA1, ANO5, ATP2A1, B3GALNT2, BAG3, BICD2, BIN1, CAPN3, CAV3, CFL2, CHKB, CNTN1, COL12A1, COL4A1, COL4A2, COL6A1, COL6A2, COL6A3, CRYAB, DES, DMD, DNAJB6, DYSF, EMD, FDX1L, FHL1, FKRP, FKTN, FLNC, GAA, GMPPB, GOLGA2, INPP5K, ISPD, ITGA7, KBTBD13, KLHL40, KLHL41, LAMA2, LARGE, LDB3, LIMS2, LMNA, LMOD3, MAP3K20, MEGF10, MICU1, MME, 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, MTM1, MYH7, MYO18B, MYOT, NEB, PABPN1, PNPLA2, POGLUT1, POLG, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PYROXD1, RBCK1, RYR1, SELENON, SEPT9, SGCA, SGCB, SGCD, SGCG, SMCHD1, SPEG, SPTBN4, SYNE1, TCAP, TMEM126B, TMEM43, TNNT1, TNPO3, TOR1AIP1, TPM2, TPM3, TRAPPC11, TRIM32, TTN, VMA21, VPS13A

10.  
Kapsamlı  
Epilepsi  
Paneli  
Genleri;

✓ ABAT, ABCA2, ABCD1, ACTL6B, ADAM22, ADAR, ADPRHL2, ADSL, AFG3L2, AGA, AIFM1, AIMP1, ALDH3A2, ALDH5A1, ALDH7A1, ALG13, ALKBH8, AMACR, AMT, ANKRD11, AP2M1, AP3B2, AP4B1, AP4E1, AP4M1, AP4S1, APOPT1, ARG1, ARHGEF9, ARID1B, ARSA, ARX, ASAH1, ASNS, ASPA, ASXL3, ATP13A2, ATP1A3, ATP6V1A, ATRX, BRAT1, BTBD, C12ORF57, CACNA1A, CACNA1B, CACNA1D, CACNA1E, CACNA1H, CACNB4, CASK, CASR, CC2D1A, CDK9, CDKL5, CERS1, CHD2, CHRNA2, CHRNA4, CHRN2, CLCN2, CLCN4, CLN3, CLN5, CLN6, CLN8, CLTC, CNKSR2, CNPY3, CNTNAP2, COA7, COL4A1, COL4A3BP, COX15, COX6B1, CPT2, CSF1R, CSNK2B, CSTB, CTC1, CTSD, CTSF, CUL4B, CYFIP2, CYP27A1, D2HGDH, DARS, DARS2, DCX, DDC, DDX3X, DEGS1, DENND5A, DEPDC5, DHFR, DHPS, DNAJC5, DNM1, DNM1L, DOCK7, DPYD, DPYS, DYRK1A, EARS2, ECHS1, ECM1, EEF1A2, EFHC1, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF3F, EPM2A, EPRS, ETFA, ETFB, ETFDH, ETHE1, FA2H, FAM126A, FAR1, FARS2, FDFT1, FDX1L, FGF12, FH, FLNA, FOLR1, FOXG1, FOXRED1, FRRS1L, FUT8, GABBR2, GABRA1, GABRB2, GABRB3, GABRG2, GALC, GAMT, GCDH, GCH1, GFAP, GFM1, GFM2, GJC2, GLB1, GLDC, GLRB, GNAO1, GNB1, GNE, GOLGA2, GOSR2, GPAA1, GPHN, GRIA3, GRIA4, GRIK2, GRIN1, GRIN2A, GRIN2B, GRN, GTPBP3, HACE1, HCN1, HCN2, HECW2, HEPACAM, HIBCH, HNRNPU, HSD17B10, HSPD1, HTRA1, HTT, IBA57, ICK, IQSEC2, IRF2BPL, KCNA1, KCNA2, KCNB1, KCNC1, KCNH1, KCNMA1, KCNQ2, KCNQ3, KCNQ5, KCNT1, KCNT2, KCTD3, KCTD7, KDM5C, KIAA2022, KIF1A, KMT2E, L2HGDH, LGII, LMNB1, LRPPRC, LYRM7, MACF1, MAGI2, MARS2, MBD5, MBOAT7, MDH2, MECP2, MED12, MED17, MEF2C, MFSDB, MIPEP, MLC1, MOCS1, MRPL44, 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, MTFMT, MTHFR, MTOR, NACC1, NBEA, NDST1, NDUFAF3, NDUFAF5, NDUFAF6, NDUFS2, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NECAP1, NEU1, NEUROD2, NFU1, NHLRC1, NKX6-2, NOTCH3, NPRL3, NRXN1, NT5C2, NUBPL, NUS1, OFD1, OPHN1, P4HTM, PARS2, PCDH19, PGK1, PHACTR1, PHF6, PIGA, PIGB, PIGC, PIGG, PIGN, PIGO, PIGP, PIGQ, PIGS, PIGT, PIGV, PIGW, PITRM1, PLAA, PLCB1, PLP1, PNKP, PNPO, POLG, POLR3A, POLR3B, PPP2CA, PPP3CA, PPT1, PRICKLE1, PRIMA1, PRODH, PROSC, PRRT2, PSAP, PTPN23, PTS, PUM1, PURA, PYCR2, QARS, QDPR, RAB39B, RALA, RARS, RELN, RMND1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF216, ROGDI, RORB, SAMHD1, SCARB2, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SCN9A, SCO1, SDHAF1, SERAC1, SERPINI1, SIK1, SLC12A5, SLC13A5, SLC19A3, SLC1A4, SLC25A1, SLC25A15, SLC25A22, SLC25A42, SLC2A1, SLC35A2, SLC39A8, SLC46A1, SLC6A1, SLC6A8, SLC9A6, SMARCA2, SMC1A, SMS, SNAP25, SNORD118, SOX10, SPATA5, SPTAN1, SPTBN4, ST3GAL3, ST3GAL5, STX1B, STXBP1, SUMF1, SUOX, SYN1, SYNGAP1, SYNJ1, SZT2, TAF1, TBC1D20, TBC1D24, TBCD, TBCE, TBCK, TBL1XR1, TCF4, TPK1, TPP1, TRAK1, TREX1, TRIM8, TSC1, TSC2, TTC19, TUBB4A, UBA5, UBE2A, UBE3A, UNC80, VAMP2, VARS, VPS13A, WARS2, WASF1, WDR26, WDR45, WWOX, YY1, ZEB2, ZFYVE26

**11. Konjenital Miyastenik Sendromlar Paneli Genleri;**

- ✓ AGRN, CHAT, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, COLQ, DOK7, DPAGT1, FLAD1, GFPT1, LAMB2, MUSK, MYO9A, PLEC, PREPL, RAPSN, SCN4A, STIM1, SYT2

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

- ✓ GAMT, GATM, SLC6A8

**13. Demans Paneli Genleri;**

- ✓ ABCA7, APOE, APP, CHMP2B, CSF1R, FUS, GRN, MAPT, 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, PRNP, PSEN1, PSEN2, RNF216, SIGMAR1, SNCA, SORL1, TARDBP, TREM2, TUBA4A, UBE3A, UBQLN2, VCP



**14. Distoni Paneli Genleri;**

- ✓ ADCY5, ANO3, ATP1A3, BCAP31, CACNA1B, CACNA1G, DCAF17, DNAJC12, FA2H, FITM2, GCH1, GNAL, KCNMA1, KMT2B, MECR, MIPEP, 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, PDE10A, PDGFB, PDGFRB, PNKD, PRKRA, PRRT2, SGCE, SLC2A1, SLC39A14, SPR, TH, THAP1, TOR1A, UBTF, VAC14

**15. Emery-Dreifuss Kas Distrofisi Paneli Genleri;**

- ✓ DMD, EMD, FHL1, LMNA, TMEM43, TTN

**16. Epileptik  
Ensefalopati  
Paneli Genleri;**

✓ ABAT, ACTL6B, ADAM22, ADAR, ADPRHL2, ADSL, ALDH7A1, ALG13, AMT, AP2M1, AP3B2, APOPT1, ARHGEF9, ARX, ASNS, ATP6V1A, BRAT1, CACNA1A, CACNA1B, CACNA1E, CASK, CDKL5, CHD2, CLCN4, CLTC, CNKSR2, CNPY3, CNTNAP2, COX6B1, CPT2, CYFIP2, D2HGDH, DCX, DENND5A, DNM1, DNMI1L, DOCK7, ECHS1, EEF1A2, ETHE1, FAR1, FARS2, FGF12, FLNA, FOXG1, FRRS1L, GABBR2, GABRA1, GABRB2, GABRB3, GABRG2, GAMT, GLDC, GNAO1, GPHN, GRIN1, GRIN2A, GRIN2B, GTPBP3, HCN1, HECW2, HEPACAM, HIBCH, HNRNPU, HTT, KCNA2, KCNB1, KCNMA1, KCNQ2, KCNQ3, KCNQ5, KCNT1, KCNT2, KCTD3, KIF1A, LRPPRC, LYRM7, MBD5, MDH2, MECP2, MED17, MEF2C, MOCS1, MRPL44, 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, MTFMT, MTHFR, NACC1, NDUFAF6, NDUFS2, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NECAP1, NEUROD2, NRXN1, NUBPL, PARS2, PCDH19, PHACTR1, PIGA, PIGB, PIGP, PIGQ, PIGS, PLAA, PLCB1, PNKP, PNPO, POLG, PPP3CA, PROSC, PTPN23, PURA, RMND1, RNASEH2A, RNASEH2B, ROGDI, SAMHD1, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SCO1, SDHAF1, SERAC1, SIK1, SLC12A5, SLC13A5, SLC19A3, SLC25A1, SLC25A22, SLC2A1, SLC35A2, SLC6A8, SLC9A6, SNAP25, SPTAN1, ST3GAL3, ST3GAL5, STXBP1, SYN1, SYNGAP1, SYNJ1, SZT2, TBC1D24, TBCD, TBCE, TBCK, TCF4, TRAK1, TREX1, TRIM8, TSC1, TSC2, TTC19, UBA5, UBE3A, UNC80, VARS, WARS2, WDR45, WWOX, ZEB2

**17.  
Holoprozensefali  
Paneli Genleri;**

- ✓ CDON, FGF8, FGFR1, FOXH1, GLI2, GLI3, NODAL, PTCH1, SHH, SIX3, TGIF1, ZIC2

**18. İdiyopatik  
Genelleştirilmiş  
ve Fokal Epilepsi  
Panelleri Genleri;**

- ✓ ALDH7A1, AMACR, CACNA1H, CACNB4, CASR, CHRNA2, CHRNA4, CHRN2, CLCN2, DEPDC5, EFHC1, GABRA1, GABRB3, GABRG2, GRIN2A, KCNA1, KCNC1, KCNQ2, KCNQ3, KCNT1, LGI1, MTOR, NPRL3, POLG, PRRT2, RELN, SCN1A, SCN1B, SCN2A, SCN8A, SCN9A, SLC2A1, SLC6A1, STX1B, TBC1D24

**19. Lökodistrofi  
ve  
Lökoensefalopati  
Paneli Genleri;**

- ✓ ABCD1, ADAR, AIFM1, AIMP1, ALDH3A2, AP4B1, AP4E1, AP4M1, AP4S1, APOPT1, ARSA, ASPA, CLCN2, COA7, COL4A1, COX15, COX6B1, CSF1R, CTC1, CYP27A1, D2HGDH, DARS, DARS2, DEGS1, EARS2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EPRS, FA2H, FAM126A, FDX1L, FOLR1, FOXRED1, GALC, GFAP, GFM1, GJC2, HEPACAM, HIBCH, HSPD1, HTRA1, IBA57, L2HGDH, LMNB1, LYRM7, MARS2, MLC1, MRPL44, 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, MTFMT, NDUFAF5, NFU1, NKX6-2, NOTCH3, NT5C2, NUBPL, PLP1, POLR3A, POLR3B, PSAP, PYCR2, RARS, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF216, SAMHD1, SCO1, SDHAF1, SERAC1, SLC1A4, SNORD118, SOX10, SUMF1, TREX1, TTC19, TUBB4A, ZFYVE26

**20. LGMD ve  
Konjenital  
Musküler Distrofi  
Paneli Genleri;**

- ✓ ANO5, B3GALNT2, BICD2, CAPN3, CAV3, CHKB, COL4A1, COL4A2, CRYAB, DES, DMD, DNAJB6, DYSF, FKRP, FKTN, GAA, GMPPB, GOLGA2, INPP5K, ISPD, ITGA7, LAMA2, LARGE, LIMS2, LMNA, MAP3K20, MEGF10, MSTO1, MYH7, MYO18B, MYOT, PNPLA2, POGlut1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PYROXD1, RYR1, SELENON, SGCA, SGCB, SGCD, SGCG, SMCHD1, SPEG, SPTBN4, SYNE1, TCAP, TNPO3, TOR1AIP1, TRAPPC11, TRIM32, TTN, VMA21

**21. Lizensefali  
Paneli Genleri;**

- ✓ ACTB, ACTG1, ARX, ATP6V0A2, CIT, CRADD, DCX, FKTN, ISPD, KATNB1, KIAA1109, LAMB1, LARGE, MACF1, PAFAH1B1, POMGNT2, POMT1, RELN, TMTC3, TUBA1A, TUBB2B, TUBG1, VLDLR, YWHAE

**22. Makrosefali /  
overgrowth  
ilişkili  
sendromlar  
Paneli Genleri;**

- ✓ AKT1, AKT3, ASPA, ASXL2, BRWD3, CCND2, CDKN1C, CHD8, CUL4B, DHCR24, DIS3L2, DNMT3A, EED, EIF2B5, EZH2, GFAP, GLI3, GPC3, GPSM2, GRIA3, HEPACAM, HUWE1, KDM1A, KIAA0196, KIF7, KPTN, L1CAM, MED12, MLC1, MPDZ, NFIB, NFIX, NSD1, OFD1, PIGA, PIK3CA, PIK3R2, PTCH1, PTEN, RAB39B, RNF135, SETD2, SYN1, TMEM94, TSC1, TSC2, UPF3B, ZBTB20

**23. Metabolik  
Epilepsi Paneli  
Genleri;**

- ✓ ABAT ,ADSL, AGA, ALDH5A1, ALDH7A1, AMT, ARG1, BTD, D2HGDH, DHFR, DNM1L, DPYD, DPYS, ETFA, ETFB, ETFDH, ETHE1, FH GAMT, GCDH, GCH1, GLDC, GNE, GPHN, HIBCH, L2HGDH, MIPPEP, MOCS1, 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, NDUFAF3, NDUFS6, PGK1, PNPO, POLG, PRODH, PTS, QDPR, SERAC1, SLC25A1, SLC25A15, SLC25A42, SLC2A1, SLC39A8, SLC46A1, SUOX, TPK1, WARS2

**24. 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

**25. Mikrosefali ve  
Pontoserebellar  
Hipoplazi Paneli  
Genleri;**

- ✓ AKT3, AMPD2, ASPM, ASXL1, ASXL3, ATR, CASK, CCDC47, CDK5RAP2, CENPF, CENPJ, CEP152, CEP63, CSNK2A1, DONSON, DYNC1H1, DYRK1A, EFTUD2, EXOSC3, GFM1, GPT2, KANSL1, KATNB1, KIF11, LIG4, MBD5, MCPH1, MED17, MFSD2A, MIPEP, MRE11A, MYCN, MYO18B, NCAPD3, NDE1, NHEJ1, OPHN1, PAFAH1B1, PCDH12, PCLO, PCNT, PHGDH, PLK4, PNKP, POMT1, PQBP1, QARS, RARS2, RTTN, SEPSECS, SLC1A4, SMARCA2, SMARCE1, SOX11, STAG2, STAMBP, STIL, TBC1D20, TBC1D23, THOC6, TMTC3, TOE1, TOP3A, TRMT10A, TSEN2, TSEN54, TUBB, TUBB2B, TUBGCP4, TUBGCP6, UBE3B, VARS, VRK1, WDR62, WDR73, XRCC4, ZNF148, ZNF335

**26. Migren Paneli  
Genleri;**

- ✓ ATP1A2 ,ATP1A3, CACNA1A, KCNK18, 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, NOTCH3, POLG, PRRT2, SCN1A, SLC1A3, SLC2A1

**27. NCL ve  
Progresif  
Miyoklonik  
Epilepsi Paneli  
Genleri;**

- ✓ AFG3L2, ASAH1, ATP13A2, BRAT1, CERS1, CLN3, CLN5, CLN6, CLN8, CSTB, CTSD, CTSF, DNAJC5, EPM2A, FARS2, FOLR1, GABRB2, GOSR2, GRN, KCNC1, KCTD7, MFSD8, NEU1, NHLRC1, POLG, PPT1, PRICKLE1, SCARB2, SERPINI1, TBC1D24, TPP1

**28. Nemaline  
Miyopati Paneli  
Genleri;**

- ✓ ACTA1, BIN1, CFL2, KBTBD13, KLHL40, KLHL41, LMOD3, MTM1, MYO18B, NEB, TNNT1, TPM2, TPM3

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

- ✓ ACO2, AFG3L2, ANTXR1, APTX, ATAD3A, AUH, C10ORF2, C12ORF65, C19ORF12, CHN1, CISD2, DNAJC19, DNM1L, 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

**30. Nöronal Göç  
Bozukluğu Paneli  
Genleri;**

- ✓ ACTB, ACTG1, ADGRG1, AKT3, ARFGEF2, ARX, ATP6V0A2, B3GALNT2, COL4A1, COL4A2, DCX, DYNC1H1, EMX2, FAT4, FH, FKTN, FLNA, FLVCR2, GMPPB, GPSM2, ISPD, KATNB1, KIF1BP, KIF7, LICAM, LAMA2, LAMB1, LAMC3, LARGE, MACF1, MED12, MEF2C, MPDZ, NDE1, NSDHL, OCLN, PAFAH1B1, PHGDH, PIK3CA, PIK3R2, POMGNT2, POMT1, RAB18, RAB3GAP1, RAB3GAP2, RELN, RTTN, SEPSECS, SRPX2, TMEM5, TUBA1A, TUBA8, TUBB2A, TUBB2B, TUBB3, TUBG1, VLDLR, WDR62, YWHAE

**31. Parkinson Hastalığı Paneli Genleri;**

- ✓ ATP13A2, ATP1A3, DNAJC6, FBXO7, GCH1, LRRK2, MAPT, 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, PARK2, PARK7, PDGFEB, PDGFRB, PINK1, PLA2G6, PRKRA, SLC20A2, SLC39A14, SLC6A3, SNCA, SPR, SYNJ1, TH, VPS13A, VPS13C, VPS35, XPR1

**32. Periyodik Felç Paneli Genleri;**

- ✓ CACNA1S, CLCN1, KCNJ2, SCN4A

**33. Polimikrogri Paneli Genleri;**

- ✓ ADGRG1, AKT3, FH, GPSM2, KIF1BP, LAMC3, NDE1, NSDHL, OCLN, PI4KA, RAB18, SNAP29, SRPX2, TBC1D20, TUBA1A, TUBA8, TUBB2A, TUBB2B, TUBB3, WDR62, ALAD, ALAS2, CPOX, FECH, HFE, HMBS, PPOX, UROD, UROS

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

- ✓ HESX1, OTX2, PAX6, SOX2



**35. Spastik  
Parapleji Paneli  
Genleri;**

- ✓ ABCD1, AFG3L2, ALDH18A1, ALS2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARG1, ARL6IP1, ATAD3A, ATLL1, B4GALNT1, BSCL2, BTBD, C12ORF65, C19ORF12, CACNA1G, CAPN1, COASY, CTNNA1, CYP27A1, CYP2U1, CYP7B1, DARS, DDHD1, DDHD2, ERLIN2, FA2H, FARS2, FXN, GALC, GBA2, GBE1, GCH1, GJC2, GPT2, HACE1, HSPD1, IBA57, IRF2BPL, KDM5C, KIAA0196, KIDINS220, KIF1A, KIF5A, L1CAM, L2HGDH, MARS2, NIPA1, NKX6-2, NT5C2, PAH, PLP1, PNPLA6, RARS, REEP1, RTN2, SACS, SETX, SLC16A2, SLC1A4, SLC25A15, SLC33A1, SPAST, SPG11, SPG20, SPG7, SPR, TECPR2, TFG, TH, UBAP1, ZFYVE26

**36. Spinal  
Muskuler Atrofi  
Paneli Genleri;**

- ✓ AARS, ASAH1, ATP7A, BICD2, BSCL2, CHCHD10, DCTN1, DNAJB2, DYNC1H1, EXOSC3, EXOSC8, FBXO38, GARS, HEXA, HSPB1, HSPB3, HSPB8, IGHMBP2, LAS1L, PLEKHG5, REEP1, SCO2, SLC5A7, SMN1, SMN2, TBCE, TRPV4, UBA1, VAPB, VPK1

**37.  
Tuberosklerozis  
Paneli Genleri;**

- ✓ TSC1, TSC2

**38. X'e Baęlı  
Entelektüel  
Yetersizlik Paneli  
Genleri;**

- ✓ ABCD1, ACSL4, AFF2, AP1S2, ARHGEF6, ARHGEF9, ARX, ATP6AP2, ATP7A, ATRX, BCOR, BRWD3, CASK, CDKL5, CLCN4, CNKSR2, CUL4B, DCX, DDX3X, DKC1, DLG3, ELK1, FANCB, FGD1, FLNA, FMR1, FRMPD4, FTSJ1, GDI1, GK, GPC3, GRIA3, HCCS, HDAC8, HPRT1, HSD17B10, HUWE1, IDS, IGBP1, IL1RAPL1, IQSEC2, KDM5C, KIAA2022, KLF8, LICAM, LAMP2, MAGT1, MAOA, MBTPS2, MECP2, MED12, MID1, MSL3, MTM1, NDP, NDUFA1, NHS, NLGN3, NLGN4X, NONO, NSDHL, NXF5, OCRL, OFD1, OPHN1, OTC, PAK3, PCDH19, PDHA1, PGK1, PHF6, PHF8, PLP1, PORCN, PQBP1, PRPS1, PTTTHOC2CHD1, RAB39B, RBM10, RLIM, RPL10, RPS6KA3, SHROOM4, SLC16A2, SLC6A8, SLC9A6, SMC1A, SMS, SOX3, SRPX2, SYN1, SYP, TAF1, TIMM8A, TSPAN7, UBE2A, UPF3B, USP9X, ZC4H2, ZCCHC12, ZDHHC9, ZNF41, ZNF674, ZNF711, ZNF81