Giltig från
2023-04-13

Centrum för Medfödda Metabola Sjukdomar

## Genpanel EP version 20 (533 gener)
## Gener kopplade till epileptisk encefalopati. 165 gener ingår i IEM-panelen v.20.

*AARS1, ABAT, ABCA2, ACTL6B, ADAM22, ADAR, ADARB1, ADAT3, ADCY5, ADGRG1, ADGRL1, ADNP, ADPRS, ADRA2B, ADSL, AFF3, AFG3L2, AGO1, AGTR2, AIMP1, AKT1, AKT3, ALDH5A1, ALDH7A1, ALG1, ALG11, ALG13, ALG14, ALG3, ALG6, ALG9, ALKBH8, AMT, ANKRD11, ANKRD17, AP1G1, AP2M1, AP3B2, AP4B1, AP4S1, APP, ARFGEF1, ARFGEF2, ARHGEF9, ARID1A, ARID1B, ARV1, ARX, ASAH1, ASH1L, ASNS, ASXL3, ATAD1, ATN1, ATP13A2, ATP1A1, ATP1A2, ATP1A3, ATP2B1, ATP5PO, ATP6AP2, ATP6V0A1, ATP6V0A2, ATP6V0C, ATP6V1A, ATP6V1B2, ATP7A, ATRX, ATXN2, BAP1, BCAS3, BCL11A, BRAF, BRAT1, BRWD3, BSCL2, CACNA1A, CACNA1B, CACNA1C, CACNA1D, CACNA1E, CACNA1G, CACNA1I, CACNA2D1, CACNA2D2, CACNG2, CAD, CAMK2A, CAMK2B, CAMLG, CASK, CCM2, CCND2, CDK19, CDKL5, CELF2, CEP85L, CERS1, CERT1, CHD2, CHD4, CHD5, CHKA, CHRNA2, CHRNA4, CHRNA7, CHRNB2, CLCN3, CLCN4, CLDN5, CLN3, CLN5, CLN6, CLN8, CLTC, CNKSR2, CNNM2, CNPY3, CNTN2, CNTNAP1, CNTNAP2, COL4A1, COL4A2, CPA6, CPLX1, CRADD, CSDE1, CSNK1E, CSNK2A1, CSNK2B, CSTB, CTSD, CTSF, CUL3, CUX2, CYFIP2, DALRD3, DCX, DDX23, DDX3X, DEAF1, DENND5A, DEPDC5, DHDDS, DLG4, DMXL2, DNAJA1, DNAJC5, DNM1, DNM1L, DNMT3A, DOCK7, DOLK, DPM1, DPM2, DPYD, DYNC1H1, DYRK1A, EEF1A2, EHMT1, EIF2S3, ELFN1, EMC10, EML1, EPM2A, FBXO28, FGF12, FGF13, FGFR3, FLNA, FOLR1, FOXG1, FOXP1, FRRS1L, FZR1, GABBR1, GABBR2, GABRA1, GABRA2, GABRA3, GABRA4, GABRA5, GABRB1, GABRB2, GABRB3, GABRD, GABRG2, GAD1, GAL, GAMT, GATM, GCSH, GET4, GLDC, GLI3, GLRA1, GLS, GLYCTK, GNAI1, GNAO1, GNB1, GNB5, GOSR2, GOT2, GPAA1, GPHN, GPT2, GRIA2, GRIA3, GRIA4, GRIK2, GRIN1, GRIN2A, GRIN2B, GRIN2D, GRM7, GRN, GUF1, HACE1, HCN1, HCN2, HEATR5B, HECW2, HID1, HIVEP2, HNRNPH2, HNRNPR, HNRNPU, HPDL, HSD17B10, IER3IP1, IQSEC2, IRF2BPL, ITPA, JMJD1C, KANSL1, KAT5, KATNB1, KCNA1, KCNA2, KCNA6, KCNB1, KCNC1, KCNC2, KCND2, KCND3, KCNH1, KCNH5, KCNJ10, KCNJ11, KCNK4, KCNMA1, KCNN2, KCNN3, KCNQ1, KCNQ2, KCNQ3, KCNQ5, KCNT1, KCNT2, KCTD3, KCTD7, KDM4B, KDM5C, KIF1A, KIF5A, KIF5C, KMT2A, KMT2E, KRIT1, LAMB1, LGI1, LIAS, LMBRD2, LMNB2, LNPK, MACF1, MADD, MAGEL2, MAPK10, MARCHF6, MAST3, MBD5, MBOAT7, MDH1, MDH2, MECP2, MED13L, MED17, MED23, MEF2C, MFF, MFSD8, MICAL1, MID2, MOCS1, MOCS2, MTHFS, MTOR, NAA10, NACC1, NAPB, NARS1, NBEA, NCDN, NDE1, NECAP1, NEDD4, NEUROD2, NEXMIF, NF1, NGLY1, NHLRC1, NPRL2, NPRL3, NR2F1, NR4A2, NRROS, NRXN1, NSF, NSRP1, NTNG2, NTRK2, NUP214, NUS1, OGDHL, OPHN1, OTUD6B, OXR1, PACS2, PAFAH1B1, PAK1, PAK3, PARS2, PAX7, PCDH12, PCDH19, PCDHGC4, PDE2A, PDK1, PGAP2, PGAP3, PHACTR1, PHF21A, PIGA, PIGB, PIGC, PIGF, PIGG, PIGH, PIGK, PIGL, PIGN, PIGO, PIGP, PIGQ, PIGS, PIGT, PIGU, PIGV, PIGW, PIGY, PIK3R2, PLCB1, PLP1, PLPBP, PLXNA1, PNKP, PNPO, POLG, PPFIBP1, PPP1CB, PPP2CA, PPP2R1A, PPP3CA, PPT1, PRDM8, PRICKLE1, PRICKLE2, PRIMA1, PRODH, PRRT2, PRUNE1, PTPN11, PTPN23, PUM1, PURA, QARS1, RAB11A, RAB11B, RAB39B, RALGAPA1, RAPGEF2, RARS2, RBSN, RELN, RFT1, RHOBTB2, RNASEH2A, RNASEH2B, RNASEH2C, RNF13, RNF2, ROGDI, RORA, RORB, RUSC2, SAMD12,*

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*SAMHD1, SATB1, SCAF4, SCARB2, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SEMA6B, SEPSECS, SERPINI1, SETBP1, SETD1A, SETD1B, SGCE, SHANK3, SIK1, SLC12A5, SLC13A5, SLC19A3, SLC1A2, SLC1A4, SLC25A12, SLC25A22, SLC2A1, SLC32A1, SLC35A2, SLC35A3, SLC38A3, SLC39A8, SLC45A1, SLC6A1, SLC6A8, SLC6A9, SLC7A6OS, SLC9A6, SMARCA2, SMC1A, SNAP25, SNIP1, SNX27, SORCS3, SPATA5, SPATA5L1, SPG11, SPTAN1, SPTBN1, SRPX2, ST3GAL3, ST3GAL5, STAMBP, STARD7, STRADA, STT3B, STX1A, STX1B, STXBP1, STYXL1, SUMF1, SV2A, SYN1, SYNGAP1, SYNJ1, SYP, SZT2, TBC1D24, TBC1D2B, TBCD, TBCK, TBL1XR1, TCF4, TDP2, THOC2, TMTC3, TNPO2, TNRC6A, TPP1, TRAK1, TRAPPC10, TRAPPC12, TRAPPC4, TRAPPC6B, TREX1, TRIM8, TRIO, TRPM3, TRPM6, TSC1, TSC2, TUBA1A, TUBB2A, TUBB2B, TUBGCP2, UBA5, UBE2A, UBE3A, UBR7, UFC1, UFSP2, UGDH, UGP2, UNC13B, UNC80, USP7, VARS1, VPS13A, WASF1, WDR26, WDR37, WDR45, WDR45B, WDR62, WWOX, YEATS2, YIF1B, YIPF5, YWHAG, ZBTB18, ZDHHC9, ZEB2, ZMYND11, ZNF142, ZNF526, ZNHIT3.*