

Epilepszia (Epilepsy) génpanel v:2022.01.26.

Panel tartalma 1681 gén:

AASS, AARS, AARS2, ABAT, ABCA2, ABCB7, ABCC8, ABCC9, ABCD1, ABCD3, ABHD16A, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACO2, ACOX1, ACP5, ACTB, ACTG1, ACTL6B, ACVR1, ACY1, ACSF3, ACSL4, ADA, ADAM22, ADAMTSL2, ADAR, ADARB1, ADAT3, ADD3, ADGRG1, ADGRV1, ADK, ADNP, ADPRHL2, ADPRS, ADRA2B, ADSL, AFF2, AFF3, AFG3L2, AGA, AGK, AGMO, AGPS, AGTR2, AHCY, AHDC1, AHI1, AIFM1, AIMP1, AIMP2, AKT1, AKT2, AKT3, ALAD, ALDH18A1, ALDH3A2, ALDH4A1, ALDH5A1, ALDH7A1, ALDOA, ALDOB, ALG1, ALG11, ALG12, ALG13, ALG14, ALG2, ALG3, ALG6, ALG8, ALG9, ALKBH8, ALMS1, ALPL, ALX4, AMACR, AMER1, AMPD2, AMT, ANK2, ANK3, ANKH, ANKLE2, ANKRD11, ANKRD17, ANO10, ANO3, ANTXR1, ANTXR2, AP1G1, AP1S2, AP2M1, AP2S1, AP3B1, AP3B2, AP3D1, AP4B1, AP4E1, AP4M1, AP4S1, APC2, APOPT1, APP, APTX, AQP2, ARF1, ARF3, ARFGEF1, ARFGEF2, ARG1, ARHGAP31, ARHGEF15, ARHGEF6, ARHGEF9, ARID1A, ARID1B, ARID2, ARL13B, ARMC9, ARNT2, ARSA, ARSB, ARV1, ARX, ASAH1, ASCL1, ASH1L, ASL, ASNS, ASPA, ASPM, ASS1, ASTN1, ASTN2, ASXL1, ASXL2, ASXL3, ATAD1, ATIC, ATM, ATN1, ATP13A2, ATP1A1, ATP1A2, ATP1A3, ATP2A2, ATP2B3, ATP5A1, ATP5F1A, ATP5F1E, ATP6AP1, ATP6AP2, ATP6V0A2, ATP6V0C, ATP6V1A, ATP6V1B2, ATP7A, ATP7B, ATP8A2, ATPAF2, ATR, ATRX, AUH, AUTS2, AVPR2, B3GALNT2, B3GLCT, B4GALNT1, B4GALT1, B4GAT1, B9D1, BAZ2B, BCAP31, BCAS3, BCKDHA, BCKDHB, BCKDK, BCL11A, BCL11B, BCOR, BCORL1, BCS1L, BDNF, BEST1, BMP4, BOLA3, BPNT2, BPTF, BRAF, BRAT1, BRPF1, BRSK2, BRWD3, BSCL2, BSN, BTB, BUB1B, C12orf57, C12orf65, C19orf12, C2CD3, C2orf69, CA5A, CA8, CACNA1A, CACNA1B, CACNA1C, CACNA1D, CACNA1E, CACNA1G, CACNA1H, CACNA1I, CACNA2D1, CACNA2D2, CACNB4, CAD, CAMK2A, CAMK2B, CAMK2G, CAMTA1, CAPN10, CARS2, CASK, CASR, CAV1, CBL, CBS, CC2D1A, CC2D2A, CCBE1, CCDC115, CCDC186, CCDC88A, CCDC88C, CCM2, CCND2, CD96, CDC42, CDC42BPB, CDH11, CDH15, CDH2, CDK10, CDK13, CDK19, CDK5, CDK5RAP2, CDK6, CDK8, CDK9, CDKL5, CDKN1C, CDON, CELF4, CENPE, CENPJ, CEP104, CEP120, CEP135, CEP152, CEP164, CEP290, CEP41, CEP57, CEP63, CEP85L, CERS1, CERT1, CHAMP1, CHAT, CHD1, CHD1L, CHD2, CHD4, CHD5, CHD7, CHD8, CHKB, CHL1, CHRM1, CHRNA2, CHRNA4, CHRNA7, CHRNB2, CIC, CILK1, CIT, CKAP2L, CLCF1, CLCN2, CLCN3, CLCN4, CLCN6, CLDN16, CLDN19, CLIP1, CLN2(TPP1), CLN3, CLN5, CLN6, CLN8, CLP1, CLPB, CLPP, CLTC, CNKSR2, CNNM2, CNOT1, CNOT3, CNPY3, CNTN2, CNTN4, CNTNAP1, CNTNAP2, COA3, COA5, COA7, COA8, COASY, COG1, COG4, COG5, COG6, COG7, COG8, COL11A2, COL18A1, COL2A1, COL4A1, COL4A2, COL4A3BP, COLGALT1, COMT, COQ2, COQ4, COQ5, COQ6, COQ8A, COQ9, COX10, COX14, COX15, COX20, COX6B1, COX7B, CP, CPA6, CPLANE1, CPLX1, CPS1, CPT1A, CPT2, CRADD, CRB2, CRBN, CREBBP, CRH, CRIPT, CRLF1, CRPPA, CTC1, CTCF, CTNNA2, CTNNB1, CTNND2, CTNS, CTSA, CTSC, CTSD, CTSF, CTSK, CTU2, CUL3, CUL4B, CUX1, CUX2, CYB5R3, CYC1, CYFIP2, CYP27A1, CYP27B1, CYP2U1, CYP7B1, CSDE1, CSF1R, CSMD1, CSNK1G1, CSNK2A1, CSNK2B, CSPP1, CSTB, D2HGDH, DAG1, DARS, DARS2, DBH, DBT, DCAF17, DCC, DCHS1, DCX, DDC, DDOST, DDX3X, DEAF1, DEGS1, DENND5A,

DEPDC5, DGUOK, DHCR24, DHCR7, DHDDS, DHFR, DHPS, DHTKD1, DHX16, DHX30, DIAPH1, DIP2B, DIP2C, DIS3L2, DISP1, DKC1, DLAT, DLD, DLG1, DLG2, DLG3, DLG4, DLGAP2, DLL1, DLL4, DMBX1, DMD, DMXL2, DNA2, DNAJC12, DNAJC19, DNAJC5, DNAJC6, DNM1, DNM1L, DNMT3A, DOCK6, DOCK7, DOLK, DPAGT1, DPH1, DPM1, DPM2, DPM3, DPYD, DPYS, DSCAM, DYM, DYNC1H1, DYRK1A, EARS2, EBF3, EBP, ECHS1, ECM1, EDC3, EEF1A2, EFHC1, EFTUD2, EGF, EHHADH, EHMT1, EIF2A, EIF2AK2, EIF2AK3, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF2S3, EIF3F, ELAC2, ELMO2, ELOVL4, ELP4, EMC1, EMC10, EMG1, EML1, EMX2, ENPP1, EOGT, EP300, EPB41L1, EPG5, EPM2A, EPRS, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, ERF, ERLIN2, ERMARD, ESCO2, ETFA, ETFB, ETFDH, ETHE1, EXOC6B, EXOC7, EXOSC3, EXOSC8, EXT2, EXTL3, EZH2, F2, F5, FA2H, FAH, FAM111A, FAM126A, FAM50A, FAR1, FARS2, FARSB, FASN, FASTKD2, FAT4, FBN1, FBP1, FBXL4, FBXO11, FBXO28, FBXO31, FDFT1, FDX1L, FDX2, FDXR, FGD1, FGF12, FGF13, FGF23, FGF8, FGFR1, FGFR2, FGFR3, FH, FHL1, FIG4, FKRP, FKTN, FLNA, FLRT3, FLVCR1, FLVCR2, FMN2, FMR1, FOLR1, FOXG1, FOXH1, FOXP1, FOXRED1, FRMPD4, FRRS1L, FTL, FTO, FTSJ1, FUCA1, FUK, FUT8, FXYP2, G6PC1, GAA, GABBR2, GABRA1, GABRA2, GABRA3, GABRA5, GABRA6, GABRB1, GABRB2, GABRB3, GABRD, GABRG2, GABRG3, GAD1, GAL, GALC, GALNS, GALNT2, GALT, GAMT, GAN, GAS1, GATA3, GATA6, GATAD2B, GATM, GBA, GBE1, GCDH, GCH1, GCSH, GDI1, GFAP, GFER, GFM1, GFM2, GFPT1, GJA1, GJB1, GJC2, GK, GLA, GLB1, GLDC, GLI2, GLI3, GLRA1, GLRA2, GLRB, GLS, GLUD1, GLUL, GLYCTK, GM2A, GMPPA, GMPPB, GNAI1, GNAO1, GNAQ, GNAS, GNB1, GNB2, GNB5, GNE, GNPAT, GNPTAB, GNPTG, GNRH1, GNS, GOLGA2, GOSR2, GOT2, GPAA1, GPC3, GPC4, GPHN, GPSM2, GPT2, GRIA2, GRIA3, GRIA4, GRIK2, GRIN1, GRIN2A, GRIN2B, GRIN2D, GRIP1, GRM1, GRM7, GRN, GSS, GTF2H5, GTF2IRD1, GTPBP2, GTPBP3, GUF1, GUSB, H3-3A, H3-3B, H3F3A, H3F3B, HACE1, HADH, HADHA, HADHB, HARS2, HAX1, HCFC1, HCN1, HCN2, HCN4, HCCS, HDAC4, HDAC8, HEATR5B, HECW2, HEPACAM, HERC1, HERC2, HESX1, HEXA, HEXB, HGSNAT, HIBCH, HID1, HIVEP2, HLCS, HMGCL, HMGCS2, HNRNPH2, HNRNPK, HNRNPR, HNRNPU, HOXA1, HPD, HPDL, HPRT1, HRAS, HSD17B10, HSD17B4, HSPD1, HTRA1, HTRA2, HTT, HUWE1, HYAL1, IARS2, IBA57, ICK, IDH2, IDH3A, IDS, IDUA, IER3IP1, IFIH1, IFT140, IKBKG, IL12B, IL1RAPL1, ILF2, INO80, INPP5E, INPP5K, INS, INSR, INTS6, IQSEC2, IRF2BP1, ISCA2, ISPD, ITPA, ITPR1, IVD, JAG1, JAKMIP1, JAM3, JMJD1C, KANK1, KANSL1, KARS, KARS1, KAT2B, KAT5, KAT6A, KAT6B, KAT8, KATNB1, KCNA1, KCNA2, KCNB1, KCNC1, KCNC3, KCND2, KCND3, KCNH1, KCNH2, KCNH5, KCNJ1, KCNJ10, KCNJ11, KCNJ2, KCNJ6, KCNK4, KCNMA1, KCNQ2, KCNQ3, KCNQ5, KCNT1, KCNT2, KCTD3, KCTD7, KDM2B, KDM3B, KDM4B, KDM5B, KDM5C, KDM6A, KDM6B, KIAA0586, KIAA1109, KIAA1715, KIAA2022, KIF11, KIF1A, KIF1BP, KIF2A, KIF4A, KIF5A, KIF5C, KIF7, KIFBP, KIRREL3, KISS1, KLHL15, KLHL7, KMT2C, KMT2D, KMT2E, KMT5B, KPNA7, KPTN, KRAS, KRIT1, L1CAM, L2HGDH, LAMA2, LAMB1, LAMC3, LAMP2, LARGE1, LARS, LARS1, LARS2, LAS1L, LBR, LDB3, LEO1, LGI1, LGI4, LIAS, LIG4, LINGO1, LIPA, LIPT1, LIPT2, LMAN2L, LMBRD1, LMBRD2, LMNB1, LMNB2, LNPB, LONP1, LRP2, LRPPRC, LSS, LZTFL1, LYRM7, LYST, MACF1, MADD, MAF, MAGEL2, MAGI2, MAGT1, MAN1B1, MAN2B1, MANBA, MAOA, MAP1B, MAP2K1, MAP2K2, MAP3K3, MAPK10, MAPK8IP3, MAPRE2, MARS2, MAST1, MAT1A, MATN4, MBD5, MBOAT7, MBTPS2, MCCC1, MCCC2, MCM3AP, MCOLN1, MCPH1, MDH2, MECP2, MED12, MED12L, MED13, MED13L, MED17, MED23, MED25,

MED27, MEF2C, MEGF10, MEIS2, METTL23, MFF, MFN2, MFSD2A, MFSD8, MGAT2, MGME1, MGP, MICAL1, MID1, MID2, MINPP1, MIPEP, MKS1, MLC1, MLPH, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MMUT, MN1, MOCS1, MOCS2, MOCS3, MOGS, MORC2, MPC1, MPDU1, MPDZ, MPI, MPLKIP, MPV17, MRAP, MRPL44, MRPS22, MSL3, MSX2, MTFMT, MTHFR, MTHFS, MTO1, MTOR, MTPAP, MTR, MTRR, MVK, MYH3, MYO1H, MYO5A, MYOT, MYT1L, NAA10, NAA15, NACC1, NADK2, NAGA, NAGLU, NAGS, NALCN, NANS, NAPB, NARS, NARS1, NARS2, NAT8L, NAXD, NAXE, NBAS, NBEA, NCDN, NCKAP1, NCOR1, NDE1, NDN, NDP, NDST1, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA2, NDUFA4, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFAF8, NDUFB11, NDUFB3, NDUFB8, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NECAP1, NEDD4L, NEU1, NEUROD2, NEXMIF, NF1, NFIA, NFIB, NFIX, NFU1, NGLY1, NHLRC1, NID1, NIN, NIPA1, NIPBL, NKX6-2, NLGN1, NLGN3, NLGN4X, NLRP3, NNT, NONO, NOTCH3, NOVA2, NPC1, NPC2, NPHP1, NPHP3, NPRL2, NPRL3, NR2F1, NR4A2, NRAS, NRROS, NRXN1, NRXN3, NSD1, NSDHL, NSF, NSUN2, NT5C2, NTNG1, NTNG2, NTRK2, NUBPL, NUP107, NUP188, NUP214, NUS1, OAT, OCLN, OCRL, OFD1, OGT, OPA1, OPA3, OPHN1, OSGEP, OTC, OTUD6B, OTUD7A, OTX2, OXR1, P4HTM, PACS1, PACS2, PAFAH1B1, PAH, PAK1, PAK3, PANK2, PARP6, PARS2, PAX6, PBX1, PC, PCCA, PCCB, PCDH12, PCDH19, PCDHB4, PCDHGC4, PCK1, PCLO, PCNT, PCYT2, PDE4D, PDE6D, PDGFRB, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PET100, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PGAP1, PGAP2, PGAP3, PGK1, PGM1, PGM2L1, PHACTR1, PHC1, PHF21A, PHF3, PHF6, PHGDH, PHYH, PI4KA, PIDD1, PIEZO2, PIGA, PIGB, PIGC, PIGG, PIGH, PIGK, PIGL, PIGM, PIGN, PIGO, PIGP, PIGQ, PIGS, PIGT, PIGU, PIGV, PIGW, PIGY, PIK3AP1, PIK3CA, PIK3R2, PITRM1, PLA2G6, PLAA, PLCB1, PLCG2, PLEKHG2, PLK4, PLP1, PLPBP, PMM2, PMPCA, PMPCB, PNKD, PNKP, PNPLA8, PNPO, PNPT1, POGZ, POLA1, POLG, POLG2, POLR2A, POLR3A, POLR3B, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POU1F1, POU3F3, PPA2, PPARG, PPIL1, PPM1D, PPM1K, PPP1R15B, PPP2CA, PPP2R1A, PPP2R5D, PPP3CA, PPT1, PQBP1, PRDM8, PREPL, PRF1, PRICKLE1, PRICKLE2, PRIMA1, PRKAR1A, PRMT7, PRODH, PROP1, PROSC, PRPS1, PRRT2, PRSS12, PRUNE, PSAP, PSAT1, PSEN1, PSMB8, PSMD12, PSPH, PTCH1, PTCHD1, PTEN, PTF1A, PTPN11, PTPN23, PTPN4, PTRH2, PTS, PUF60, PUM1, PURA, PUS1, PUS3, PYCR2, QARS, QARS1, QDPR, QRIC1, RAB11A, RAB11B, RAB18, RAB27A, RAB39B, RAB3GAP1, RAB3GAP2, RAC1, RAD21, RAF1, RAI1, RALA, RALGAPA1, RALGAPB, RANBP2, RARS, RARS1, RARS2, RBBP8, RBFOX1, RBFOX3, RBM10, RBM8A, RBPJ, RECQL4, REEP1, RELN, RERE, RFT1, RHOBTB2, RIT1, RMND1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF113A, RNF125, RNF13, RNF168, RNF2, RNF216, RNU4ATAC, ROGDI, RORA, RORB, RPGRIP1L, RPIA, RPL10, RPS6KA3, RRM2B, RSRC1, RTN4IP1, RTTN, RUBCN, RUSC2, RXYLT1, RYR2, RYR3, SACS, SAMHD1, SARS, SASS6, SATB1, SATB2, SC5D, SCAF4, SCAMP5, SCARB2, SCN10A, SCN1A, SCN1B, SCN2A, SCN2B, SCN3A, SCN5A, SCN8A, SCN9A, SCO1, SCO2, SCP2, SCYL2, SDCCAG8, SDHA, SDHAF1, SDHB, SDHD, SEC23B, SEC24D, SEC31A, SEMA6B, SEPSECS, SERAC1, SERPINI1, SET, SETBP1, SETD1A, SETD1B, SETD2, SETD5, SFXN4, SGCE, SGSH, SH2B1, SHANK1, SHANK3, SHH, SHROOM4, SIK1, SIN3A, SIX3, SKI, SLC12A1, SLC12A3, SLC12A5, SLC12A6, SLC13A5, SLC16A1, SLC16A2, SLC17A5, SLC18A2, SLC19A2, SLC19A3, SLC1A1, SLC1A2, SLC1A3,

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Forrás:

Genetic epilepsy syndromes (Version 2.487)

<https://panelapp.genomicsengland.co.uk/panels/402/> (Megtekintve: 2022.01.26.)

Invitae Epilepsy Panel

<https://www.invitae.com/en/physician/tests/03401/> (Megtekintve: 2022.01.26.)

Epilepsy panel

<https://www.centoportal.com/order/new/test-catalog/analysis-method?search=epilepsy>

(Megtekintve: 2022.01.26.)

Comprehensive Epilepsy Panel

<https://blueprintgenetics.com/tests/panels/neurology/comprehensive-epilepsy-panel/>

(Megtekintve: 2022.01.26.)

Comprehensive Epilepsy and Seizure Panel

<https://www.preventiongenetics.com/testInfo?val=Comprehensive+Epilepsy+and+Seizure+Panel>

(Megtekintve: 2022.01.26.)