

Gen-Panels Epilepsien

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Hinweise:

- Gen-Panels mit mehr als 10 Genen dürfen gemäss Analysenliste des BAG nur durch Ärzte mit einem FMH-Titel Medizinische Genetik verordnet werden.
- Die Gen-Panels sind nicht endgültig und können je nach Fragestellung auf Wunsch angepasst werden.
- Die Gen-Panels können abhängig von der aktuellen Datenlage zum Zeitpunkt der Analysen von denjenigen auf dieser Liste abweichen.

Epilepsien und epileptische Enzephalopathien, umfassendes Panel v8 (708 Gene)

AARS1, ABAT, ABCA2, ACOX1, ACTL6B, ADAM22, ADAR, ADARB1, ADAT3, ADD1, ADGRG1, ADPRHL2, ADSL, AFF3, AIMP1, AIMP2, AKT3, ALDH5A1, ALDH7A1, ALG1, ALG11, ALG13, ALG14, ALG3, ALG6, ALG8, ALG9, ALKBH8, ALPL, AMPD2, AMT, ANKRD11, AP1G1, AP2M1, AP3B2, APC2, ARF1, ARF3, ARFGEF1, ARFGEF2, ARG1, ARHGEF9, ARID1B, ARV1, ARX, ASAH1, ASH1L, ASNS, ASPA, ASXL3, ATN1, ATP1A1, ATP1A2, ATP1A3, ATP2B1, ATP5A1, ATP6AP2, ATP6V0A1, ATP6V0A2, ATP6V0C, ATP6V1A, ATP7A, ATRX, BAP1, BCKDHA, BCKDHB, BCS1L, BLOC1S1, BOLA3, BRAF, BRAT1, BSCL2, BSN, BTBD, C12orf57, CACNA1A, CACNA1B, CACNA1C, CACNA1D, CACNA1E, CACNA1G, CACNA1H, CACNA1I, CACNA2D1, CACNA2D2, CACNB4, CAD, CAPRIN1, CAR3, CASK, CC2D2A, CCDC88A, CCDC88C, CDC42BPB, CDK19, CDKL5, CELF2, CEP85L, CERS1, CHD2, CHD4, CHD5, CHKA, CHRNA2, CHRNA4, CHRNB2, CIC, CLCN3, CLCN4, CLN3, CLN6, CLN8, CLPB, CLTC, CNKSR2, CNNM2, CNPY3, CNTNAP2, COG4, COG6, COG7, COG8, COL18A1, COL4A1, COL4A2, COLGALT1, COQ2, COQ4, COQ6, COQ9, COX10, COX15, CPA6, CPLX1, CPSF3, CREBBP, CSNK1G1, CSNK2B, CSTB, CTNNA2, CTSD, CTSF, CTU2, CUL3, CUL4B, CUX2, CYFIP2, CYP27A1, D2HGDH, DBT, DCX, DDC, DDX3X, DEAF1, DEGS1, DENND5A, DEPDC5, DHCR24, DHCR7, DHDDS, DHPS, DHX16, DHX30, DIAPH1, DLL1, DMXL2, DNAJC5, DNAJC6, DNM1, DNM1L, DOCK7, DOLK, DPAGT1, DPH5, DPM1, DPM2, DPYD, DROSHA, DTYMK, DYNC1H1, DYRK1A, EARS2, EEF1A2, EFHC1, EFTUD2, EHMT1, EIF2AK2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF2S3, EIF3F, EMC10, EML1, EMX2, EPG5, EPM2A, ETHE1, EXOC7, EXOSC3, EXT2, FAM50A, FAR1, FARS2, FASTKD2, FBXL4, FBXO11, FBXO28, FDFT1, FGF12, FGF13, FGFR3, FH, FKBP, FKTN, FLNA, FOLR1, FOXG1, FOXRED1, FRMD5, FRRS1L, FUCA1, FUK, FUT8, GABBR2, GABRA1, GABRA2, GABRA5, GABRB1, GABRB2, GABRB3, GABRD, GABRG2, GAD1, GALC, GALNT2, GAMT, GBA, GCH1, GCSH, GFAP, GFM1, GLB1, GLDC, GLI3, GLRA1, GLRA2, GLS, GLUD1, GLUL, GLYCTK, GM2A, GNAO1, GNAQ, GNB1, GNB5, GOSR2, GOT2, GPAA1, GPHN, GRIA2, GRIA4, GRIK2, GRIN1, GRIN2A, GRIN2B, GRIN2D, GRN, GSS, GTPBP2, GTPBP3, GUF1, H3F3A, H3F3B, HACE1, HAX1, HCCS, HCFC1, HCN1, HCN2, HEATR5B, HECW2, HEPACAM, HERC2, HEXA, HEXB, HID1, HLCS, HMGCL, HNRNPH2, HNRNPR, HNRNPU, HOXA1, HPDL, HPRT1, HRAS, HSD17B4, HSPD1, HTRA2, IER3IP1, IFIH1, IKBKG, IQSEC2, IRF2BP1, ISPD, ITPA, JAKMIP1, KARS1, KAT5, KAT8, KATNB1, KCNA1, KCNA2, KCNB1, KCNC1, KCNC2, KCND2, KCNH1, KCNJ10, KCNJ11, KCNK4, KCNMA1, KCNQ2, KCNQ3, KCNQ5, KCNT1, KCNT2, KCTD3, KCTD7, KIAA1109, KIF1A, KIF1BP, KIF2A, KIF5C, KLHL20, KMT2E, KPTN, KRAS, LARGE1, LARS1, LGI1, LIAS, LIPT1, LIPT2, LMAN2L, LMBRD2, LMNB1, LNPBK, LSS, LYST, MACF1, MADD, MAF, MAGI2, MANBA, MAP2K1, MAP2K2, MAST1, MBD5, MBOAT7, MDH2, MECP2, MED12, MED17, MED27, MEF2C, MFF, MFSD8, MINPP1, MLC1, MMACHC, MMADHC, MOCS1, MOCS2, MOGS, MPDU1, MTHFR, MTHFS, MTOR, NACC1, NAGA, NAPB, NARS1, NARS2, NBEA, NCDN, NDE1, NDP, NDUFA1, NDUFA10, NDUFA2, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFS1, NDUFS2, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NECAP1, NEDD4L, NEUROD2, NEXMIF, NGLY1, NHLRC1, NPRL2, NPRL3, NR4A2, NRROS, NRXN1, NSD1, NSDHL, NSF, NSRP1, NTRK2, NUBPL, NUP214, NUS1, OCLN, OGDHL, OPHN1, OTUD6B, OTX2, OXR1, P4HTM, PABPC1, PACS1, PACS2, PAFAH1B1, PAH, PAK1, PARP6, PARS2, PCCA, PCCB, PCDH12, PCDH19, PCDHGC4, PCLO, PCYT2, PDHA1, PDHX, PDSS2, PET100, PEX1, PEX10, PEX12, PEX13, PEX19, PEX2, PEX3, PEX5, PEX6, PEX7, PGM2L1, PHACTR1, PHGDH, PIDD1, PIGA, PIGB, PIGC, PIGG, PIGH, PIGK, PIGN, PIGO, PIGP, PIGQ, PIGS, PIGT, PIGU, PIGW, PIK3R2, PLA2G6, PLAA, PLCB1, PLPBP, PMM2, PMPCB, PNKP, PNPO, PNPT1, POLG, POLG2, POMGNT1, POMT1, POMT2, PPIL1, PPP2CA, PPP3CA, PPT1, PRICKLE1, PRMT7, PRODH, PRPF8, PRRT2, PSAP, PSAT1, PSPH, PTEN, PTF1A, PTPN23, PTS, PUM1, PURA, QARS1, QDPR, RAB11A, RAB11B, RAB18, RAB3GAP1, RAB3GAP2, RALA, RALGAP1, RANBP2, RARS1, RARS2, RELN, RFT1, RHOTB2, RMND1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF113A, RNF13, RNF2, RNU4ATAC, ROGDI, RORA, RORB, RPIA, RRM2B, RTN4IP1, RTTN, RUSC2, RYR2, SAMHD1, SARS1, SATB1, SATB2, SCAF4, SCAMP5, SCARB2, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SCO1, SCO2, SDHA, SEMA6B, SEPSECS, SERPINI1, SETBP1, SETD1A, SETD1B, SETD5, SGSH, SIK1, SIX3, SLC12A5, SLC13A5, SLC16A2, SLC1A2, SLC1A4, SLC25A1, SLC25A12, SLC25A22, SLC2A1, SLC31A1, SLC35A1, SLC35A2, SLC35A3, SLC38A3, SLC39A8, SLC45A1, SLC5A6, SLC6A1, SLC6A8, SLC9A6, SMARCA2, SMARCC2, SMC1A, SMS, SNAP25, SNIP1, SNORD118, SNX27, SPATA5, SPATA5L1, SPR, SPTAN1, SPTBN1, SPTBN4, ST3GAL3, ST3GAL5, STAG1, STAMBIP, STARD7, STRADA, STX1B, STXBP1, SUCLA2, SUCLG1, SUOX, SURF1, SYN1, SYNCRIP, SYNGAP1, SYNJ1, SZT2, TAF8, TANC2, TANGO2, TBC1D20, TBC1D24, TBC1D2B, TBCD, TBCK, TBL1XR1, TCF4, TDP2, Telo2, TET3, TFE3,

TIAM1, TIMM50, TMEM106B, TMEM222, TMEM70, TMX2, TNK2, TNPO2, TPP1, TRAF7, TRAK1, TRAPPC12, TRAPPC4, TRAPPC6B, TREX1, TRIM8, TRIP13, TRPM3, TRPM6, TRRAP, TSC1, TSC2, TSEN15, TSEN2, TSEN34, TSEN54, TSFM, TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, TUBGCP2, TXNRD1, UBA5, UBE2A, UBE3A, UBR7, UFC1, UFM1, UFSP2, UGDH, UGP2, UNC80, USP18, USP7, VAMP2, VARS1, VLDLR, VPS11, VPS50, WARS2, WASF1, WDR37, WDR45, WDR45B, WDR62, WDR73, WNK3, WWOX, XK, YIF1B, YIPF5, YWHAG, ZBTB18, ZDHHC9, ZEB2, ZMIZ1, ZMYM2, ZNF142, ZNF335

Benigne familiäre neonatale / infantile Epilepsie v5 (6 Gene)

ATP1A2, KCNQ2, KCNQ3, PRRT2, SCN2A, SCN8A

Familiäre hemiplegische Migräne v6 (15 Gene)

ATP1A2, ATP1A3, CACNA1A, COL4A1, COL4A2, CSNK1D, KCNK18, NOTCH3, PNKD, PRRT2, SCN1A, SLC1A3, SLC2A1, SLC4A4, TREX1

Hyperekplexie v5 (18 Gene)

ARHGEF9, ASNS, ATAD1, CACNA1A, CLPB, CRLF1, CTNNB1, GLRA1, GLRB, GPHN, HEXA, RPS6KA3, SCN8A, SLC6A5, SLC6A9, SUOX, TRAK1, TSEN54

Idiopathische generalisierte und fokale Epilepsie v5 (65 Gene)

ALDH7A1, AP2M1, ARFGEF1, ATP1A2, ATP1A3, CACNA1A, CACNA1C, CACNA1H, CACNB4, CERS1, CHD2, CHRNA2, CHRNA4, CHRN2, CPA6, DEPDC5, DNM1, EFHC1, EPM2A, FGF13, GABRA1, GABRB3, GABRD, GABRG2, GNAO1, GRIN2A, GRIN2B, HCN1, HCN2, HNRNPU, KCNA1, KCNA2, KCNC1, KCND2, KCNMA1, KCNQ2, KCNQ3, KCNT1, KCTD7, LGI1, MTOR, NHLRC1, NPRL2, NPRL3, PCDH19, PIGH, POLG, PRRT2, RELN, RORB, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SIK1, SLC12A5, SLC2A1, SLC6A1, STX1B, STXBP1, SYN1, SYNGAP1, TBC1D24, TXNRD1