

## Epilepsie und Migräne

**Dravet-Syndrom [GP016] (~21 kb)**

1. *SCN1A*, 2. *GABRG2*, *SCN2A*, *SCN9A*, *STXBP1*

**Epilepsie [GP017] (~25 kb)**

1. *SCN1A*, 2. *CDKL5*, *GABRA1*, *GABRG2*, *KCNQ2*, *PCDH19*, *STXBP1*, *SYNGAP1*

**erweiterte Diagnostik [GP017XL]**

*ACY1*, *ADSL*, *ALDH7A1*, *AMT*, *ARHGEF9*, *ARX*, *CACNA1H*, *CACNB4*, *CDKL5*, *CHD2*, *CHRNA2*, *CHRNA4*, *CHRN2*, *CLCN2*, *CNTNAP2*, *CPA6*, *CPT2*, *EFHC1*, *EPM2A*, *FOLR1*, *FOXP1*, *GABRA1*, *GABRB3*, *GABRD*, *GABRG2*, *GAMT*, *GCSH*, *GLDC*, *GRIN2A*, *GRIN2B*, *JRK*, *KCNJ10*, *KCNMA1*, *KCNQ2*, *KCNQ3*, *KCNT1*, *LGI1*, *MAGI2*, *MAPK10*, *MECP2*, *MEF2C*, *MTHFR*, *NRXN1*, *PCDH19*, *PLCB1*, *PNKP*, *PNPO*, *PRRT2*, *RNASEH2A*, *RNASEH2B*, *RNASEH2C*, *SAMHD1*, *SCN1A*, *SCN1B*, *SCN2A*, *SCN8A*, *SCN9A*, *SLC19A3*, *SLC25A22*, *SLC2A1*, *SLC9A6*, *SPTAN1*, *SRPX2*, *STXBP1*, *SYNGAP1*, *TBCE*, *TCF4*, *TREX1*, *UBE3A*, *ZEB2*

**Absence-Epilepsie der Kindheit, CAE [GP015] (~14 kb)**

*CACNA1H*, *GABRA1*, *GABRB3*, *GABRG2*, *JRK*, *SLC2A1*

**Frühinfantile Epileptische Enzephalopathie (EIEE; Ohtahara-Syndrom) [GP019XL]**

*ARHGEF9*, *ARX*, *CDKL5*, *KCNQ2*, *PCDH19*, *PLCB1*, *PNKP*, *SCN1A*, *SCN2A*, *SCN8A*, *SLC25A22*, *SPTAN1*, *STXBP1*

**Epileptische Enzephalopathie [GP018] (~24kb)**

*CDKL5*, *GABRA1*, *SCN1A*, *SCN2A*, *SCN8A*, *STXBP1*

**erweiterte Diagnostik [GP018XL]**

*ACY1*, *ADAR*, *ADSL*, *ALDH7A1*, *ALG13*, *AMT*, *ARHGEF9*, *ARX*, *BRAT1*, *CACNA1A*, *CASK*, *CDKL5*, *CHD2*, *CNTNAP2*, *CPT2*, *DCX*, *DNM1*, *FLNA*, *FOLR1*, *FOXP1*, *GABRA1*, *GABRB3*, *GABRG2*, *GAMT*, *GCSH*, *GLDC*, *GPHN*, *GRIN1*, *GRIN2A*, *GRIN2B*, *HDAC4*, *HNRNPU*, *KCNJ10*, *KCNQ2*, *MAGI2*, *MAPK10*, *MBD5*, *MECP2*, *MEF2C*, *MOCS1*, *MOCS2*, *MTHFR*, *NRXN1*, *PCDH19*, *PLCB1*, *PNKP*, *PNPO*, *PRRT2*, *RNASEH2A*, *RNASEH2B*, *RNASEH2C*, *ROGDI*, *SAMHD1*, *SCN1A*, *SCN1B*, *SCN2A*, *SCN8A*, *SCN9A*, *SLC19A3*, *SLC25A22*, *SLC2A1*, *SLC9A6*, *SPTAN1*, *ST3GAL3*, *ST3GAL5*, *STXBP1*, *SYN1*, *SYNGAP1*, *TBC1D24*, *TBCE*, *TCF4*, *TREX1*, *TSC1*, *TSC2*, *UBE3A*, *WDR45*, *ZEB2*

**Generalisierte Epilepsie mit Fieberkrämpfen plus (GEFS+) [GP020] (~21kb)**

1. *SCN1A* 2. *GABRD*, *GABRG2*, *SCN1B*, *SCN2A*, *SCN9A*

**MELAS-Syndrom [GP164]**

*MT-TL1*, *MT-ND1*, *MT-ND5*, *MT-ND4*, *MT-ND6*, *MT-CYB*, *MT-CO3*, *MT-CO1*, *MT-CO2*, *MT-ND3*, *MT-TF*, *MT-TQ*, *MT-TH*, *MT-TV*, *MT-TW*, *MT-TK*, *MT-TL2*, *MT-TE*, *MT-TC*, *MT-TS1*, *MT-TS2*

**Hyperekplexie, hereditäre [GP021] (~9 kb)**

*ARHGEF9*, *GLRA1*, *GPRB*, *GPHN*, *SLC6A5*

**Metabolische Epilepsie [GP022XL]**

*ACY1*, *ADSL*, *AGA*, *ALDH4A1*, *ALDH5A1*, *ALDH7A1*, *AMT*, *ARG1*, *ATIC*, *BTD*, *DPYD*, *ETFA*, *ETFB*, *ETFDH*, *FH*, *FOLR1*, *GAMT*, *GCDH*, *GCH1*, *GCSH*, *GLDC*, *GNE*, *GPHN*, *HPD*, *L2HGDH*, *MOCS1*, *MOCS2*, *MTHFR*, *PCBD1*, *PGK1*, *PNPO*, *PRODH*, *PTS*, *QDPR*, *SLC25A15*, *SLC46A1*, *SUOX*

**Nächtliche Frontallappenepilepsie, autosomal dominante (ADNFLE) [GP177] (~14kb)**

*CRH*, *CHRNA2*, *CHRNA4*, *CHRN2*, *DEPDC5*, *KCNT1*

**Familiäre hemiplegische Migräne (FHM) [GP023] (~23 kb)**

*ATP1A2*, *ATP1A3*, *CACNA1A*, *SCN1A*