

## Developmental and epileptic encephalopathy génpanel v:2025-05-22

Panel tartalma 116 gén:

AARS1, ACTL6B, ADAM22, ALG13, AP3B2, ARHGEF9, ARV1, ARX, ATP1A2, ATP1A3, ATP6V0A1, ATP6V1A, CACNA1A, CACNA1E, CACNA2D1, CAD, CDK19, CDKL5, CELF2, CHD2, CNPY3, CPLX1, CUX2, CYFIP2, DALRD3, DENND5A, DEPDC5, DMXL2, DNMT1, DOCK7, EEF1A2, FBXO28, FGF12, FGF13, FRRS1L, FZR1, GABBR2, GABRA1, GABRA2, GABRA5, GABRB1, GABRB2, GABRB3, GABRG2, GAD1, GLS, GLUL, GNAO1, GOT2, GRIN1, GRIN2B, GRIN2D, GUF1, HCN1, HID1, HNRNPU, ITPA, KCNA2, KCNB1, KCNC2, KCNH5, KCNQ2, KCNT1, KCNT2, MAST3, MDH1, MDH2, NAPB, NECAP1, NEUROD2, NSF, NTRK2, PACS2, PARS2, PCDH19, PHACTR1, PIGA, PIGB, PIGP, PIGQ, PIGS, PLCB1, PNKP, PPP3CA, RHOBTB2, RNF13, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SIK1, SLC12A5, SLC13A5, SLC1A2, SLC25A12, SLC25A22, SLC32A1, SLC35A2, SLC38A3, SMC1A, SNF8, SPTAN1, ST3GAL3, STXBP1, SV2A, SYNJ1, SZT2, TBC1D24, TRAK1, UBA5, UFSP2, UGDH, UGP2, WWOX, YWHAG

Forrás:

Developmental and epileptic encephalopathy - PS308350 Phenotypic Series (2025-04-14)

<https://www.omim.org/phenotypicSeries/PS308350>

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