

Epilepsie

Epilepsie volledig genpanel

344

EPI00v24.1

Gennaam	CreV4 (15X)	SSv7 (15X)
RORA	100	100
CLN6	100	99.8
AP3B2	100	100
POLG	100	100
CHD2	100	99.8
NPRL3	100	100
PIGQ	100	100
TSC2	100	100
TBC1D24	100	100
ATP6V0C	100	100
ROGDI	100	99.5
ABAT	100	100
GRIN2A	100	100
NDE1	100	100
CLN3	100	99.2
PRRT2	100	100
SETD1A	99.9	99.1
STX1B	100	96.4
KAT8	100	100
SLC6A2	100	100
GNAO1	100	100
GOT2	100	99.4
AARS	100	100
WWOX	100	100
GCSH	81.6	99.4
ANKRD11	100	100
PAFAH1B1	100	94.7
SLC13A5	100	100
MPDU1	100	100
RAI1	100	100
PIGW	100	100
NEUROD2	100	100
TUBG1	100	100
GRN	100	100
KANSL1	100	100
GOSR2	100	100
PNPO	100	100
CACNA1G	100	99.9
CLTC	100	100
TANC2	100	100
STRADA	100	100
ASXL3	100	100
NEDD4L	100	98
PIGN	100	98.6

Gennaam CreV4 (15X) SSv7 (15X)

HCN2	78.8	77.1
GAMT	100	100
SEMA6B	100	99.3
DHPS	100	100
RNASEH2A	100	100
NACC1	100	100
CACNA1A	100	100
MAST3	99.9	98.4
CERS1	95.4	78.3
SCN1B	99.9	93.8
ATP1A3	100	100
GRIK5	100	100
CIC	100	100
KPTN	100	100
GRIN2D	96.5	81
PNKP	100	100
MBOAT7	100	100
ITPA	100	100
PLCB1	100	99.9
SNAP25	100	100
NAPB	100	100
PIGU	100	99.7
SAMHD1	100	100
PIGT	100	100
SLC12A5	100	99
KCNB1	100	100
DPM1	100	100
CHRNA4	100	96.4
KCNQ2	100	97.6
EEF1A2	99.6	97.5
DNAJC5	100	100
USP25	99.9	99.4
SCAF4	100	98
SYNJ1	100	97.7
SON	100	100
PIGP	100	100
DYRK1A	100	100
SIK1	99.9	100
CSTB	100	100
SLC25A1	99.9	94.5
CLDN5	100	100
TBX1	91.5	84.3
TANGO2	100	100
DEPDC5	100	100
ADSL	100	100
MLC1	100	100
SHANK3	95.9	90.2
CLCN4	100	100
OFD1	100	99.1
PIGA	100	100

Gennaam CreV4 (15X) SSv7 (15X)

CDKL5	100	98.6
RPS6KA3	99.8	98.9
CNKSR2	100	99.4
SMS	99.8	100
EIF2S3	100	100
ARX	93.1	88
ATP6AP2	100	100
DDX3X	100	99.9
CASK	100	98.3
SYN1	100	100
PQBP1	100	100
SLC35A2	100	99.6
WDR45	100	100
SYP	100	97
KDM5C	100	100
IQSEC2	99.6	98.9
SMC1A	100	100
HSD17B10	100	100
HUWE1	100	99.9
FGD1	100	99.8
ARHGEF9	100	100
OPHN1	100	100
MED12	100	99.9
KIAA2022	100	100
ATRX	100	98.8
PCDH19	100	100
HNRNPH2	100	100
PLP1	100	100
PAK3	100	99.8
DCX	100	99.9
ALG13	100	99.7
UBE2A	100	100
CUL4B	100	98.2
GRIA3	100	99.9
ZDHHC9	100	100
GPC3	100	100
PHF6	100	97.6
SLC9A6	100	99.9
FGF13	100	99.9
GABRA3	100	99.9
NSDHL	100	100
SLC6A8	97.6	98.9
HCFC1	100	100
MECP2	100	99.9
FLNA	100	99.6
RAB39B	100	100
GNB1	100	100
GABRD	97.1	93.8
CHD5	99.9	97.8
MTOR	100	100

Gennaam CreV4 (15X) Ssv7 (15X)

MTHFR	100	100
HNRNPR	100	100
DHDDS	100	100
ADPRHL2	99.9	100
PPT1	100	100
SLC2A1	100	100
SZT2	100	99.9
ST3GAL3	100	100
CPT2	100	99.7
PARS2	100	100
DOCK7	100	99.1
KCNA2	100	100
KCND3	100	100
ATP1A1	100	100
PHGDH	100	100
CHRNA2	100	100
ASH1L	100	100
KCNJ10	100	100
ATP1A2	100	100
PIGC	100	100
CACNA1E	100	100
KCNT2	100	97.2
KCNH1	100	100
KCTD3	100	99.7
ARV1	100	100
TBCE	100	100
HNRNPU	100	98.7
CAD	100	100
NRXN1	99.1	100
UGP2	100	100
SLC1A4	100	100
STAMBP	100	100
ST3GAL5	100	92.1
RANBP2	99.4	98.9
ZEB2	100	100
MBD5	100	100
KIF5C	100	99.4
NR4A2	100	100
SCN3A	100	100
SCN2A	100	99.7
SCN1A	100	100
GAD1	100	100
SLC25A12	100	100
HECW2	100	100
PGAP1	100	96.9
SLC19A3	100	100
KIF1A	100	100
D2HGDH	100	100
GRM7	100	100
SLC6A1	100	99.7

Gennaam CreV4 (15X) SSv7 (15X)

GLB1	100	100
TRAK1	100	100
TREX1	100	100
QARS	100	100
AMT	100	100
NPRL2	100	100
CACNA2D2	99.7	96.9
CACNA1D	100	100
ATP6V1A	100	99.9
UBA5	100	99.5
SERPINI1	100	100
AP2M1	100	100
FGF12	100	100
PIGG	100	100
CPLX1	99.9	94.6
LIAS	100	96.3
UGDH	100	100
GABRA2	100	100
GABRB1	100	100
SCARB2	100	100
COQ2	100	99.6
PPP3CA	100	100
TBCK	100	96
SPATA5	100	100
MFSD8	100	100
GLRB	100	94.5
GRIA2	100	99.7
TRIO	99.2	99
SLC1A3	100	100
HCN1	100	100
MOCS2	100	100
MEF2C	100	100
ALDH7A1	100	100
PPP2CA	100	100
PURA	100	100
DIAPH1	99	99.7
GLRA1	100	100
GEMIN5	100	100
CYFIP2	100	100
GABRB2	100	100
GABRA1	100	100
GABRG2	92.8	100
TUBB2A	100	100
TUBB2B	100	100
FARS2	100	100
PHACTR1	100	99.7
NHLRC1	100	100
TDP2	100	100
CSNK2B	100	100
NEU1	100	100

Gennaam CreV4 (15X) Ssv7 (15X)

SYNGAP1	99.2	97.8
MOCS1	100	100
CNPY3	100	99.3
KCNQ5	100	99.3
HACE1	100	99.7
MICAL1	100	100
CDK19	100	99.9
NUS1	99.7	99.8
ARG1	100	100
EPM2A	89.7	85.1
ARID1B	99.3	98.6
BRAT1	100	100
PSPH	100	99.1
KCTD7	100	100
MDH2	100	99.9
YWHAG	100	100
ASNS	100	100
KPNA7	100	100
ACTL6B	100	100
RELN	100	100
CNTNAP2	100	100
CLN8	100	100
ASAH1	100	99.4
RHOBTB2	100	100
CHRNA2	100	100
PROSC	100	100
OTUD6B	100	99.9
INTS8	100	97.7
KCNQ3	100	99.6
GPAA1	100	100
SMARCA2	99.1	98
GLDC	100	100
PIGO	100	100
TRPM3	100	100
RORB	100	100
TRPM6	100	99.4
PSAT1	100	100
NTRK2	100	100
GABBR2	98.2	98.3
FRRS1L	99.9	86.1
STXBP1	100	100
DNM1	100	97
COQ4	100	98.9
SPTAN1	100	100
TSC1	100	100
KCNT1	99.9	99
GRIN1	100	100
EHMT1	99	98.8
CACNA1B	99.3	98.3
KCNMA1	100	99.9

Gennaam CreV4 (15X) Ssv7 (15X)

ATAD1	100	99.3
LGI1	100	100
TRIM8	100	100
CNNM2	100	100
TCF7L2	100	99.2
DEAF1	98.6	93.8
SLC25A22	100	100
CTSD	100	100
TPP1	100	100
EIF3F	100	100
DENND5A	100	100
KCNC1	100	100
SLC1A2	100	100
PHF21A	100	99.7
RNASEH2C	100	100
PACS1	99.7	94.9
FOLR1	100	100
PAK1	100	100
GRIA4	100	99.9
KMT2A	99.6	99.6
KCNA1	100	100
C12orf57	100	100
GRIN2B	100	100
PPFIBP1	100	100
DNM1L	100	100
TUBA1A	100	100
SCN8A	100	100
KCNC2	100	100
ANO4	100	100
CUX2	100	98.7
SETD1B	97.9	97.4
NBEA	99.6	98.9
UFM1	100	64.2
RNASEH2B	100	100
CLN5	100	100
FOXG1	98	99.2
KCNH5	100	99.9
GPHN	100	100
PIGH	100	100
IRF2BPL	98.7	99.2
DYNC1H1	100	100
PACS2	99.3	97.7
NPAP1	100	99.9
UBE3A	100	100
GABRB3	100	100
GABRA5	100	100
GATM	100	100
DMXL2	100	99.9
GNB5	100	100
PIGB	100	95.6