Targeted Genes and Methodology Details for Comprehensive Epilepsy With or Without Encephalopathy Gene Panel

The following applies to EPPAN / Comprehensive Epilepsy Gene Panel. Testing is performed to evaluate for the presence of variants in coding regions and extending to +/- 10 base pairs of adjacent intronic sequence on either side of the coding exons of the genes analyzed. In addition, the analysis will cover select non-coding variants. Next-generation sequencing and/or a polymerase chain reaction-based quantitative method is performed to test for the presence of copy number variants (CNV) in the genes analyzed. A polymerase chain reaction-based assay is performed to test for the presence of CSTB dodecamer repeat expansions in addition to sequence variants and deletions/duplications. Confirmation of select reportable variants may be performed by alternate methodologies based on internal laboratory criteria.

This document is intended to highlight additional evaluations for variants of high clinical interest as well as technical limitations. However, this document does not comprehensively reflect all genomic regions covered by this test. For questions regarding transcripts, or genes or regions covered, contact the laboratory at 800-533-1710.

Genomic Build: GRCh37 (hg19) unless otherwise specified

Gene	Reference Transcript[a]	Additional Evaluations	Technical Limitations
ABAT	NM_020686.6	-	-
ACO2	NM_001098.3	-	-
ACY1	NM_000666.3	-	-
ADARB1	NM_015833.4	-	CNV analysis in Exon 8 will not be performed
ADGRG1	NM_005682.7	c.-153-2228 to c.-153-2178	-
ADSL	NM_000026.4	c.-54 to c.-44	-
AFG3L2	NM_006796.3	-	-
AIFM1	NM_004208.4	-	-
AKT2	NM_001626.6	-	-
ALDH3A2	NM_000382.3	-	-
ALDH5A1	NM_001080.3	-	-
ALDH7A1	NM_001182.5	-	-
ALG13	NM_001099922.3	-	-
AMT	NM_000481.4	-	-
AP2M1	NM_004068.4	-	-
APOPT1 (COA8)	NM_032374.4	-	-
ARFGEF2	NM_006420.3	-	-
ARHGEF9	NM_015185.3	-	-
ARX	NM_139058.3	-	-
ASAH1	NM_177924.5	-	CNV analysis in Exon 4 will not be performed
ASNS	NM_133436.3	-	-
ATN1	NM_001007026.2	-	-
ATP1A2	NM_000702.4	-	-
ATP1A3	NM_152296.5	-	-
ATRX	NM_000489.5	-	-
BCKDK	NM_005881.4	-	-
BCS1L	NM_004328.5	-	-

Targeted Genes and Methodology Details for Comprehensive Epilepsy With or Without Encephalopathy Gene Panel (continued)

Gene	Reference Transcript[a]	Additional Evaluations	Technical Limitations
BOLA3	NM_212552.3	-	-
BRAT1	NM_152743.4	-	-
C12orf57	NM_138425.4	-	-
CACNA1A	NM_001127221.1	-	-
CACNA1E	NM_000721.4	-	-
CACNA2D2	NM_006030.4	-	-
CAD	NM_004341.5	-	-
CARS2	NM_024537.4	-	-
CASK	NM_003688.3	-	CNV analysis in Exon 16 will not be performed
CASK	NM_001126055.2	-	CNV analysis in Exon 15 will not be performed
CCM2	NM_031443.3	-	-
CDKL5	NM_003159.2	-	CNV analysis in Exon 19 will not be performed
CDKL5	NM_001323289.2	c.-162-7 to c.-159	-
CHD2	NM_001271.4	-	-
CHRNA2	NM_000742.4	-	-
CHRNA4	NM_000744.6	-	-
CHRNB2	NM_000748.3	-	-
CLCN4	NM_001830.4	-	-
CLN3	NM_001042432.1	-	-
CLN5	NM_006493.4	-	-
CLN6	NM_017882.3	-	-
CLN8	NM_018941.4	-	-
CNTNAP2	NM_014141.6	-	-
COA8 (APOPT1)	NM_032374.4	-	-
COG7	NM_153603.4	-	-
COG8	NM_032382.4	-	-
COL18A1	ENST00000400337.6	-	CNV analysis in Exon 1 will not be performed
COL4A1	NM_001845.6	-	-
COQ2	NM_015697.8	-	-
COQ4	NM_016035.5	-	-
COQ6	NM_182476.3	-	-
COQ8A	NM_020247.5	-	-
COQ9	NM_020312.4	-	-
COX10	NM_001303.4	-	CNV analysis in Exon 6 will not be performed
COX15	NM_004376.7	-	CNV analysis in Exon 9 will not be performed
CPT2	NM_000098.3	-	-

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Gene	Reference Transcript[a]	Additional Evaluations	Technical Limitations
CSF1R	NM_005211.3	c.1969+126 to c.1969+105	-
CSTB	NM_000100.3	A polymerase chain reaction-based assay is utilized to detect CSTB dodecamer repeat expansions upstream of the gene region. c.168+32 to c.168+11	-
CTSD	NM_001909.5	-	-
CTSF	NM_003793.4	-	-
CUL4B	NM_003588.3	-	-
D2HGDH	NM_152783.5	-	-
DCX	NM_178153.3	-	-
DDC	NM_000790.4	-	-
DDX3X	NM_001193416.3	-	-
DEPDC5	NM_001242896.3	-	-
DHFR	NM_000791.4	-	CNV analysis in Exon 6 will not be performed
DIAPH1	NM_005219.5	-	-
DLD	NM_000108.5	-	-
DMXL2	NM_001174116.2	-	-
DNAJC5	NM_025219.3	-	-
DNM1	NM_004408.4	-	-
DNM1L	NM_012062.5	-	-
DOCK7	NM_001271999.1	-	-
DYRK1A	NM_001396.4	-	-
EARS2	NM_001083614.2	-	-
EEF1A2	NM_001958.5	-	-
EHMT1	NM_024757.5	-	CNV analysis in Exon 1 will not be performed
EIF2AK2	NM_001135651.3	-	-
EPM2A	NM_005670.4	-	-
ETHE1	NM_014297.5	-	-
FARS2	NM_006567.5	-	-
FASTKD2	NM_014929.3	-	-
FBP1	NM_000507.4	-	-
FBXL4	NM_012160.4	-	-
FH	NM_000143.3	-	-
FKRP	NM_024301.5	-	-
FKTN	NM_001079802.1	c.648-1248 to c.648-1238	-
FLNA	NM_001456.3	-	-
FOLR1	NM_016725.3	-	-
FOXG1	NM_005249.5	-	-
FOXRED1	NM_017547.4	-	-

Targeted Genes and Methodology Details for Comprehensive Epilepsy With or Without Encephalopathy Gene Panel (continued)

Gene	Reference Transcript[a]	Additional Evaluations	Technical Limitations
FRRS1L	NM_014334.3	-	-
GABBR2	NM_005458.8	-	-
GABRA1	NM_000806.5	-	-
GABRB2	NM_021911.2	-	CNV analysis in Exon 10 will not be performed
GABRB3	NM_000814.6	-	-
GABRG2	NM_000816.3	-	-
GAMT	NM_000156.6	-	-
GATM	NM_001482.3	-	-
GCK	NM_000162.5	-	-
GFM1	NM_024996.5	-	-
GLDC	NM_000170.2	-	-
GLRA1	NM_000171.4	-	-
GLUL	NM_002065.6	-	-
GNAO1	NM_020988.3	-	CNV analysis in Exon 2 will not be performed
GOSR2	NM_004287.4	-	CNV analysis in Exon 2 will not be performed
GPAA1	NM_003801.4	-	-
GPC3	NM_004484.4	-	-
GPHN	NM_020806.4	-	-
GRIA3	NM_000828.4	-	-
GRIN1	NM_007327.4	-	-
GRIN2A	NM_000833.5	-	-
GRIN2B	NM_000834.4	-	-
GYS2	NM_021957.4	-	-
HCFC1	NM_005334.3	-	-
HCN1	NM_021072.4	-	-
HIBCH	NM_014362.4	-	-
HNRNPU	NM_031844.3	-	-
HSD17B10	NM_004493.3	-	-
IARS2	NM_018060.4	-	-
IBA57	NM_001010867.4	-	-
IDH2	NM_002168.3	-	-
IER3IP1	NM_016097.5	-	-
IQSEC2	NM_001111125.3	-	-
ITPA	NM_033453.4	-	-
KANSL1	NM_001193466.2	-	CNV analysis in Exon 2, Exon 3 will not be performed
KCNA1	NM_000217.3	-	-

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Gene	Reference Transcript[a]	Additional Evaluations	Technical Limitations
KCNA2	NM_004974.4	-	-
KCNB1	NM_004975.4	-	-
KCNC1	NM_001112741.1	-	-
KCNH1	NM_172362.3	-	-
KCNJ10	NM_002241.5	-	-
KCNMA1	NM_002247.4	-	-
KCNMA1	NM_001271520.2	-	-
KCNQ2	NM_172107.4	-	-
KCNQ3	NM_004519.4	-	-
KCNT1	NM_020822.3	-	-
KCTD7	NM_153033.4	-	-
KDM5C	NM_004187.4	-	-
KDM6A	NM_021140.3	-	-
KRIT1	NM_194456.1	-	-
L2HGDH	NM_024884.3	-	CNV analysis in Exon 6 will not be performed
LAMA2	NM_000426.3	-	-
LARGE1	NM_004737.6	-	-
LGI1	NM_005097.4	-	-
LIAS	NM_006859.4	-	-
LRPPRC	NM_133259.4	-	CNV analysis in Exon 19, Exon 36, Exon 38 will not be performed
MBD5	NM_018328.4	-	-
MECP2	NM_004992.3	-	-
MECP2	NM_001110792.2	-	-
MEF2C	NM_002397.5	-	-
MFSD8	NM_152778.3	-	-
MICU1	NM_006077.3	-	-
MOCS1	NM_005943.5	c.*7+10 to c.*7+1	-
MOCS2	NM_176806.4	-	-
MOCS2	NM_004531.5	-	-
MTFMT	NM_139242.4	-	-
MTO1	NM_012123.4	-	-
MTOR	NM_004958.4	-	-
NALCN	NM_052867.4	-	-
NDUFA1	NM_004541.4	-	-
NDUFA2	NM_002488.4	-	-
NDUFAF2	NM_174889.5	-	-
NDUFAF3	NM_199069.2	-	-
NDUFAF4	NM_014165.4	-	-

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Gene	Reference Transcript[a]	Additional Evaluations	Technical Limitations
NDUFAF5	NM_024120.5	-	-
NDUFAF6	NM_152416.4	c.420+779 to c.420+789	-
NDUFS1	NM_005006.7	-	-
NDUFS4	NM_002495.4	-	-
NDUFS6	NM_004553.5	-	-
NDUFS7	NM_024407.5	-	-
NDUFS8	NM_002496.4	-	-
NDUFV1	NM_007103.4	-	-
NECAP1	NM_015509.4	-	-
NEDD4L	NM_015277.6	-	CNV analysis in Exon 4, Exon 26 will not be performed
NEU1	NM_000434.4	-	-
NEXMIF	NM_001008537.3	-	-
NGLY1	NM_018297.4	-	-
NHLRC1	NM_198586.3	-	-
NOTCH3	NM_000435.3	-	-
NPRL2	NM_006545.5	-	-
NPRL3	NM_001077350.3 [GRCh38(hg38)]	-	-
NR2F1	NM_005654.6	-	-
NR4A2	NM_006186.4	-	-
NRROS	NM_198565.3	-	-
NRXN1	NM_001135659.2	-	CNV analysis in Exon 6, Exon 13 will not be performed
OCLN	NM_002538.4	-	Sequencing and CNV analysis in Exon 5 - Exon 9 will not be performed
OFD1	NM_003611.3	-	-
OPHN1	NM_002547.3	-	-
OTUD6B	NM_016023.5	-	-
P4HTM	NM_177938.2	-	-
PACS1	NM_018026.4	-	-
PACS2	NM_001100913.3	-	-
PAFAH1B1	NM_000430.4	-	CNV analysis in Exon 4 will not be performed
PAK3	NM_002578.5	-	CNV analysis in Exon 11 will not be performed
PCDH12	NM_016580.3	-	-
PCDH19	NM_001184880.2	-	-
PDCD10	NM_145860.1	-	-
PDHA1	NM_000284.4	c.292-28 to c.292-18	-
PDHB	NM_000925.4	-	-

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Gene	Reference Transcript[a]	Additional Evaluations	Technical Limitations
PDHX	NM_003477.3	-	-
PDP1	NM_018444.4	-	-
PDSS2	NM_020381.4	-	CNV analysis in Exon 7 will not be performed
PEX7	NM_000288.4	c.-50 to c.-40	-
PHF6	NM_032458.3	-	-
PHGDH	NM_006623.4	-	-
PIGA	NM_002641.3	-	-
PIGG	NM_001127178.3	-	-
PIGK	NM_005482.3	-	-
PIGL	NM_004278.4	-	-
PIGM	NM_145167.3	c.-265 to c.-275	-
PIGN	NM_176787.5	-	CNV analysis in Exon 14 will not be performed
PIGO	NM_032634.4	-	-
PIGQ	NM_004204.4	-	-
PIGS	NM_033198.4	-	-
PIGT	NM_015937.6	-	-
PIGU	NM_080476.4	-	-
PIGV	NM_017837.3	-	-
PIGW	NM_178517.4	-	-
PLCB1	NM_015192.4	-	-
PLP1	NM_000533.5	c.453+16 to c.453+54; c.453+154 to c.453+169; c.454-327 to c.454-307	-
PLPBP	NM_007198.4	-	-
PNKP	NM_007254.4	c.1387-23 to c.1386+39	-
PNPLA8	NM_015723.5	-	-
PNPO	NM_018129.4	-	-
POLG	NM_002693.2	-	-
POMGNT1	NM_017739.3	-	-
POMT1	NM_007171.3	-	-
POMT2	NM_013382.5	c.1333-11 to c.1333-19	-
PPP2R5D	NM_006245.4	-	-
PPT1	NM_000310.3	-	-
PRRT2	NM_145239.3	-	-
PURA	NM_005859.5	-	-
QARS1	NM_005051.3	-	-
RAB39B	NM_171998.4	-	-
RAB3GAP1	NM_012233.3	-	-

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Gene	Reference Transcript[a]	Additional Evaluations	Technical Limitations
RALA	NM_005402.4	-	-
RALGAPA1	NM_014990.3	-	CNV analysis in Exon 4, Exon 5, Exon 24, Exon 33 will not be performed
RANBP2	NM_006267.5	-	Sequenicng and CNV analysis in Exon 2 - Exon 20 will not be performed
RARS2	NM_020320.5	-	-
RELN	NM_005045.4	-	CNV analysis in Exon 37, Exon 64 will not be performed
RMND1	NM_017909.4	-	-
ROGDI	NM_024589.2	-	-
RRM2B	NM_015713.5	-	-
SATB2	NM_015265.4	-	-
SCARB2	NM_005506.4	-	-
SCN1A	NM_001165963.3	c.4002+2508 to c.4002+2158; c.265-2138 to c.265-2148	-
SCN1B	NM_001037.5	-	CNV analysis in Exon 1 will not be performed
SCN2A	NM_021007.3	-	-
SCN2A	NM_001040143.2	-	-
SCN3A	NM_006922.4	-	-
SCN8A	NM_014191.4	-	-
SCN8A	NM_001330260.2	-	-
SCO2	NM_005138.2	-	-
SDHAF1	NM_001042631.2	-	-
SERAC1	NM_032861.4	-	CNV analysis in Exon 3 will not be performed
SERPINI1	NM_005025.4	-	-
SETBP1	NM_015559.3	-	-
SETD2	NM_014159.6	-	CNV analysis in Exon 2 will not be performed
SIK1	NM_173354.5	-	-
SLC12A5	NM_020708.5	-	-
SLC13A5	NM_177550.5	-	-
SLC16A1	NM_003051.3	-	-
SLC19A3	NM_025243.4	c.980-11 to c.980-19	-
SLC25A1	NM_005984.5	-	-
SLC25A12	NM_003705.5	-	-
SLC25A22	NM_024698.6	-	-
SLC2A1	NM_006516.3	c.680-11 to c.680-16	-
SLC35A2	NM_001042498.3	-	-

Targeted Genes and Methodology Details for Comprehensive Epilepsy With or Without Encephalopathy Gene Panel (continued)

Gene	Reference Transcript[a]	Additional Evaluations	Technical Limitations
SLC35A3	NM_012243.3	-	CNV analysis in Exon 6 will not be performed
SLC6A1	NM_003042.4	-	-
SLC6A8	NM_005629.4	-	-
SLC9A6	NM_006359.3	-	-
SLC9A6	NM_001042537.1	-	-
SMARCA2	NM_003070.5	-	-
SMC1A	NM_006306.4	-	-
SMS	NM_004595.5	-	-
SNAP25	NM_003081.4	-	-
SNAP25	NM_130811.3	-	-
SNAP29	NM_004782.4	-	-
SNX27	NM_030918.6	-	-
SPATA5	NM_145207.3	-	-
SPR	NM_003124.5	-	-
SPTAN1	NM_001130438.3	-	-
ST3GAL3	NM_006279.5	-	-
ST3GAL5	NM_003896.4	-	-
STRADA	NM_001003787.4	-	-
STX1B	NM_052874.5	-	-
STXBP1	NM_003165.4	-	-
STXBP1	NM_001032221.4	-	-
SUCLA2	NM_003850.2	-	-
SUOX	NM_000456.3	-	-
SYN1	NM_133499.2	-	-
SYNGAP1	NM_006772.3	-	CNV analysis in Exon 14 will not be performed
SYNJ1	NM_003895.3	-	-
SYP	NM_003179.2	-	-
SZT2	NM_015284.4	-	-
TBC1D24	NM_001199107.2	-	-
TBL1XR1	NM_024665.6	-	-
TCF4	NM_001083962.2	-	-
TPK1	NM_022445.4	-	-
TPP1	NM_000391.4	-	-
TSC1	NM_000368.5	-	-
TSC2	NM_000548.5	c.848+276 to c.848+286; c.976-20 to c.976-11; c.2838-127 to c.2838-117; c.5069-22 to c.5069-13; c.5259+11 to c.5259+29	-

Targeted Genes and Methodology Details for Comprehensive Epilepsy With or Without Encephalopathy Gene Panel (continued)

Gene	Reference Transcript[a]	Additional Evaluations	Technical Limitations
TSFM	NM_001172696.2	-	CNV analysis in Exon 5 will not be performed
TUBA1A	NM_006009.4	-	-
TUBA8	NM_018943.3	-	-
TUBB2B	NM_178012.5	-	CNV analysis in Exon 4 will not be performed
TWNK	NM_021830.5	-	-
UBE3A	NM_130838.4	-	-
UGP2	NM_006759.3	-	-
USP7	NM_003470.3	-	-
VARS2	NM_001167734.1	-	-
VLDLR	NM_003383.5	-	-
WDR26	NM_025160.6	-	-
WDR37	NM_014023.4	-	-
WDR45	NM_007075.3	-	-
WDR62	NM_001083961.2	-	-
WWOX	NM_016373.4	-	-
YWHAG	NM_012479.4	-	-
ZDHHC9	NM_016032.4	-	-
ZEB2	NM_014795.4	-	-