

NEW TEST

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Comprehensive Epilepsy Panel, Sequencing and Deletion/Duplication

3001591, EPI NGS

Specimen Requirements:

Patient Preparation:

Collect: Lavender or pink (EDTA) or yellow (ACD solution A or B).

Specimen Preparation: Transport 3 mL whole blood. (Min: 2 mL)

Transport Temperature: Refrigerated.

Unacceptable Conditions: Serum or plasma; grossly hemolyzed or frozen specimens; saliva, buccal brush, or swab; FFPE tissue; DNA

Remarks:

Stability: Ambient: 72 hours; Refrigerated: 1 week; Frozen: Unacceptable

Methodology: Massively Parallel Sequencing

Performed: Varies

Reported: 14-21 days

Note: Genes Tested: AARS; ABAT*; ADGRG1; ADSL*; ALDH5A1; ALDH7A1; ALG1*; ALG13*; ALG3; ALG6; ALG8; ALG9*; AMACR; AMT; ANKRD11*; AP3B2*; ARFGEF2; ARG1; ARHGEF9*; ARV1*; ARX*; ASAH1*; ASNS; ATN1; ATP1A1; ATP1A3; ATP6AP2; ATP7A; ATRX*; BCKDK; BRAT1*; BTD*; C12orf57; CACNA1A; CACNA1D; CACNA1E; CACNA2D2; CAD; CARS2*; CASK; CDKL5; CHD2; CHRNA4; CHRN2; CLCN4; CLN3; CLN5*; CLN6*; CLN8; CLTC; CNKSR2*; CNTNAP2; COL4A1; CPT2; CSTB; CTSD; CTSF; CUL4B*; DCX; DDX3X*; DEAF1*; DEPDC5; DHDDS; DIAPH1; DMXL2*; DNAJC5; DNM1*; DNM1L; DOCK7; DPAGT1; DPM1; DPYD; DYNC1H1**; DYRK1A; EEF1A2; EHMT1*; EPM2A***; FARS2**; FGF12; FKTN*; FLNA; FOLR1; FOXP1*; FRRS1L; GABBR2*; GABRA1; GABRB2; GABRB3*; GABRD; GABRG2*; GALC; GAMT; GATM; GFAP; GNAO1; GNB1; GOSR2; GPHN*; GRIA3; GRIN1; GRIN2A; GRIN2B; HACE1; HCN1; HECW2; HNRNPU; HSD17B10; IQSEC2; ITPA; KANSL1*; KCNA1; KCNA2; KCNB1; KCNC1; KCNH1; KCNJ10; KCNJ11; KCNMA1; KCNQ2*; KCNQ3; KCNT1; KCTD7*; KDM5C*; KIF1A*; LGI1; MBD5*; MDH2; MECP2; MED17; MEF2C; MFSD8; MOCS2; MOGS; MPDU1; MTHFR; MTOR; NDE1; NECAP1; NEDD4L; NEU1; NEXMIF; NGLY1; NHLRC1; NPRL2; NPRL3; NR2F1*; NRXN1*;

NSD1; NTRK2*; OPHN1; PACS1; PAFAH1B1*; PCDH19; PEX1; PEX12; PEX2; PEX3; PEX6; PHF6; PHGDH; PIGA; PIGG; PIGN; PIGO; PIGQ; PIGT; PIGV; PLCB1; PLPBP*; PMM2; PNKP; PNPO; POLG; PPT1; PRICKLE2; PRRT2; PSAP; PTPN23; PURA; QARS1; QDPR; RELN; RFT1; RNASEH2A; RNASEH2B; RNASEH2C; ROGDI; RORB*; SAMHD1*; SATB2; SCARB2; SCN1A*; SCN1B; SCN2A; SCN3A; SCN8A; SERPINI1; SETBP1; SLC12A5; SLC13A5; SLC19A3***; SLC1A2; SLC25A12*; SLC25A22; SLC2A1; SLC35A2; SLC6A1; SLC9A6*; SMARCA2*; SMC1A; SMS; SNAP25; SPATA5; SPTAN1*; ST3GAL3*; ST3GAL5; STRADA; STX1B; STXBP1*; SUOX; SYN1; SYNGAP1*; SYNJ1; SZT2*; TBC1D24; TBL1XR1; TCF4; TPK1*; TPP1; TREX1; TSC1; TSC2; TSEN54*; UBA5; UBE3A*; UNC80*; VPS13A; WDR45; WWOX**; ZEB2* *One or more exons are not covered by sequencing and/or deletion/duplication analysis for the indicated gene; see limitations section below.
**Deletion/duplication detection is not available for this gene.
***One or more exons are not covered by sequencing, and deletion/duplication detection is not available for this gene; see limitations section below.

CPT Codes:	81419
New York DOH Approval Status:	Specimens from New York clients will be sent out to a New York DOH approved laboratory, if possible.
Interpretive Data:	
Refer to report.	
Reference Interval:	
By report	

HOTLINE NOTE: Refer to the Hotline Test Mix for interface build information.