In-solution hybridization of all coding exons is performed on the patient's genomic DNA. Although some deep intronic regions may also be analyzed, this assay is not meant to interrogate most promoter regions, deep intronic regions, or other regulatory elements, and does not detect single or multi-exon deletions or duplications. Direct sequencing of the captured regions is performed using next generation sequencing. The patient's gene sequences are then compared to a standard reference sequence. Potentially causative variants and areas of low detection significance. Variants of unknown significance may require further studies of the patient and/or family members.

The Epilepsy and Seizure Disorders Panel is comprised of a next generation sequencing (NGS) for syndromic and non-syndromic causes of seizures. It is recommended that individuals with seizures have a chromosomal microarray as a first tier test. Please click here for information on our EmArray Cyto and CytoScan SNP Array.

Support for the development of this panel was provided, in part, by a grant from the Epilepsy Foundation to Dr. Andrew Escayg, Associate Professor, Department of Human Genetics.

References:

Genes

ABAT, ADGRG1, ADSL, AKT3, ALDHA5A, ALDH7A1, ALG13, ANKR1D11, ARHGEF9, ARX, ASPH, ASXL1, ATP1A2, ATP1A3, ATP6AP2, BCKDK, CACNA1A, CACNA1C, CACNA2D2, CACNB4, CASK, CASR, CDKL5, CENPJ, CHD2, CHRNA2, CHRNB2, CLCN4, CLN3, CLN5, CLN6, CLN8, CNTNAP2, CPA6, CSTB, CTSD, CYP27A1, DCX, DEPDC5, DNASC5, DNMT1, DUOX1, EEF1A2, EFHC1, EHTM1, EPM2A, FLNA, FOLR1, FOXL1, GABRA1, GABRB1, GABRB2, GABRG2, GART, GATM, GNAO1, GOSR1, GRIN1, GRIN2A, GRIN2B, HCN1, HCN4, HNRNPU, IQSEC2, KANSL1, KCNA1, KCNA2, KCNB1, KCNC1, KCNQ1, KCNQ2, KCNQ3, KCNQ4, KCNQ5, KLC1, KCTD7, LG11, LIAS, MAGI1, MBDS, MEF2C, MEF2E, MFSD8, MTHFR, MTOR, NDE1, NDUFA1, NEDD4L1, NEXM1, NLRRC1, NPR2, NRP1, NRP2, OPN1L, PAG1, PAFH1B1, PCDH19, PHF6, PIGA, PIK3CA, PLCB1, PNKP, PNPO, POLG, PTEN, PRKCE1, PRKCE2, PRRT2, PURA, QRARS, RELN, SCARB2, SCN1A, SCN1B, SCN2A, SCN2B, SCN8A, SHH, SIK1, SIX3, SLC13A5, SLC19A3, SLC25A19, SLC25A22, SLC2A1, SLC35A2, SLC6A4, SLC9A5, SMC1A, SNAP25, SPTAN1, STAC3, STGAL3, STGAL5, STIL, STX1B, STXBP1, SYN1, SYNGAP1, SNT2, TBC1D24, TCP4, TDP1, TSC1, TSC2, TSE1M4, UBES1A, USP5X, WDR45, WDR62, WWOX, ZEB2

Indications

This test is indicated for:
- Individuals with epilepsy.

Methodology

Next Generation Sequencing: In-solution hybridization of all coding exons is performed on the patient's genomic DNA. Although some deep intronic regions may also be analyzed, this assay is not meant to interrogate most promoter regions, deep intronic regions, or other regulatory elements, and does not detect single or multi-exon deletions or duplications. Direct sequencing of the captured regions is performed using next generation sequencing. The patient's gene sequences are then compared to a standard reference sequence. Potentially causative variants and areas of low coverage are Sanger-sequenced. Sequence variations are classified as pathogenic, likely pathogenic, benign, likely benign, or variants of unknown significance. Variants of unknown significance may require further studies of the patient and/or family members.

Detection
Next Generation Sequencing: Clinical Sensitivity: Unknown. Mutations in the promoter region, some mutations in the introns and other regulatory
element mutations cannot be detected by this analysis. Large deletions/duplications will not be detected by this analysis. Results of molecular analysis
should be interpreted in the context of the patient's clinical/biochemical phenotype.

Analytical Sensitivity: ~99%.

**Specimen Requirements**

Submit only 1 of the following specimen types

**Type: Whole Blood**

Specimen Requirements:

In EDTA (purple top) tube:
- Infants (2 years): 3-5 ml
- Older Children & Adults: 5-10 ml.

Specimen Collection and Shipping: Ship sample at room temperature with overnight delivery.

**Type: Isolated DNA**

Specimen Requirements:

In microtainer: 60 ug

Isolation using the Qiagen™ Puregene kit for DNA extraction is recommended.

Specimen Collection and Shipping: Refrigerate until time of shipment in 100 ng/ul of TE buffer. Ship sample at room temperature with overnight
delivery.

**Related Tests**

- CytoScan + SNP and EmArray Cyto.
- Custom diagnostic mutation analysis (KM) is available to family members if mutations are identified by targeted mutation testing or sequencing
  analysis.
- Prenatal testing is available only for known familial mutations to individuals who are confirmed carriers of mutations. Please contact the
  laboratory genetic counselor to discuss appropriate testing prior to collecting a prenatal specimen.
- Epilepsy and Seizure Disorders: Deletion/Duplication Panel.