
Lab Dept: **Anatomic Pathology**

Test Name: **COMPREHENSIVE EPILEPSY PANEL**

General Information

Lab Order Codes: CEPL

Synonyms: Sequence Analysis and Exon-Level Deletion/Duplication Analysis of 70 Genes.

CPT Codes: 81405 x2 – Molecular Pathology procedure, level 6
81406 x4 – Molecular Pathology procedure, level 7
81407 x2 – Molecular Pathology procedure, level 8

Test Includes: Comprehensive Epilepsy Panel Gene list (Sequence Analysis and Exon-level Deletion/Duplication testing): ADSL, ALDH7A1, ALF13, ARHGEF9, ARX, ATP1A2, ATP6AP2, CACNA1A, CDKL5, CH2D, CHRNA2, CHRNA4, CHRNA7, CHRN2, CLN3, CLN5, CLN6, CLN8, CNTNAP2, CSTB, CTSD, DNAJC5, DNM1, DYRK1A, EEF1A2, EPM2A, FOLR1, FOXG1, GABRA1, GABRB2, GABRB3, GABRG2, GAMT, GATM, GOSR2, GRIN1, GRIN2A, GRIN2B, IQSEC2, KANSL1, KCNB1, KCNJ10, KCNQ2, KCNQ3, KCNT1, KCTD7, LGI1, MAGI2, MBD5, MECP2, MEF2C, MFSD8, NHLRC1, NR2F1, NRXN1, PCDH19, PIGA, PIGO, PIGV, POLG, PPT1, PRICKLE1, PRRT2, QARS, SCARB2, SCN1A, SCN1B, SCN2A, SCN8A, SLC13A5, SLC25A22, SLC2A1, SLC6A8, SLC9A6, SPTAN1, STXBP1, TBC1D24, TCF4, TPP1(CLN2), TSC1, TSC2, UBE3A, WDR45, WWOX, ZEB2

Logistics

Test Indications: To assist with decisions about treatment and management of individuals with epilepsy. Epilepsy is defined by the occurrence of at least two unprovoked seizures occurring more than 24 hours apart. It can be caused by genetic disorders, metabolic diseases, trauma, infection, and structural brain abnormalities.

The Comprehensive Epilepsy panel at GeneDx includes genes causing Mendelian forms of epilepsy. Many of these genes encode subunits of ion channels involved in stabilizing or propagating neuronal activity, including components of the voltage-gated sodium, potassium, and calcium channels and the ligand-gated gamma-aminobutyric (GABA) and nicotinic acetylcholine receptor channels. The panel also includes non-ion channel genes associated with a variety of neurotransmitter, storage, and other neurometabolic disorders, as well as genes causing syndromic forms of epilepsy, many of which are involved in transcriptional activation or repression.

Lab Testing Sections: Anatomic Pathology - Sendouts

Referred to: GeneDx (GeneDx Test: 523)

Phone Numbers: MIN Lab: 612-813-6280

STP Lab: 651-220-6550

Test Availability: 24 hours, daily

Turnaround Time: 4 weeks

Special Instructions: N/A

Specimen

Specimen Type: Whole blood

Container: Lavender (EDTA) top tube

Draw Volume: Adults: 8-10 mL
Children: 4 mL
Infants: 2 mL

Processed Volume: **Do Not** process or centrifuge whole blood.
Submit 2-5 mL EDTA whole blood in original container.

Collection: Routine blood collection

Special Processing: Lab Staff: Store unprocessed blood at room temperature. Ship at room temperature via overnight courier, using a cool pack in hot weather. If shipment is delayed, the specimen should be refrigerated for up to 7 days prior to shipping.

Patient Preparation: None

Sample Rejection: Mislabeled or unlabeled specimen

Interpretive

Reference Range: An interpretive report will be provided

Critical Values: N/A

Limitations: Neither sequencing nor exon-level array CGH can reliably detect mosaicism and/or chromosomal aberrations. Deletions/insertions of less than 250 bp are not reliably detected by array CGH. Some small sections of a few genes in this panel may have inherent sequence properties that yield suboptimal data and mutations in these regions may not be reliably detected.

Methodology: Next-gen Sequencing and Exon-Level Targeted array CGH.

References:

[GeneDx](#) April 2018

Updates:

8/25/2015:CPT update.

4/16/2018: CPT and gene list update.