About EGL Genetics

EGL Genetics specializes in genetic diagnostic testing, with nearly 50 years of clinical experience and board-certified laboratory directors and genetic counselors reporting out cases. EGL Genetics offers a combined 1000 molecular genetics, biochemical genetics, and cytogenetics tests under one roof and custom testing for all medically relevant genes, for domestic and international clients.

Equally important to improving patient care through quality genetic testing is the contribution EGL Genetics makes back to the scientific and medical communities. EGL Genetics is one of only a few clinical diagnostic laboratories to openly share data with the NCBI freely available public database ClinVar (>35,000 variants on >1700 genes) and is also the only laboratory with a free online database (EmVClass), featuring a variant classification search and report request interface, which facilitates rapid interactive curation and reporting of variants.

Epilepsy

Epilepsy is a clinically heterogeneous group of neurologic disorders affecting ~1% of the population and is defined as a condition in which an individual has recurrent and unprovoked seizures. Epilepsy’s clinical presentation is variable, with a range of seizures types and frequencies.

The etiology of epileptic disorders is as diverse as their clinical presentation; however, 40% are thought to be of genetic origin. Approximately 9% of idiopathic epilepsy is associated with rare pathogenic copy number variations within the genome, while approximately 2% of isolated epilepsy cases are monogenic in origin. The remainder of genetic cases is believed to be the result of multifactorial genetic and environmental causes.

Recommended Testing Strategy

EGL Genetics recommends performing copy number variant (CNV) analysis as a first step in epilepsy genetic testing. The CytoScan SNP Array performs a high resolution analysis to detect chromosome CNVs and uniparental disomy. Most genetic causes of epilepsy can be detected by this array, making it a cost-effective option to pursue.

Should the array test results come back negative, EGL Genetics recommends ordering its Epilepsy and Other Seizure Disorders next generation sequencing Panel. Included in this panel is testing for 109 genes, which have been been associated with syndromic and non-syndromic epilepsy, and gene-targeted aCGH deletion/duplication analysis. Please see the reverse side for a table of genes analyzed.

References:

For more information about EGL Genetics:
CALL 470.378.2200
WEB eglgenetics.com
### Epilepsy and Other Seizure Disorders

**Genes Included on Epilepsy and Other Seizure Disorders Panel***

<table>
<thead>
<tr>
<th>Test Code</th>
<th>Test Name</th>
<th>CPT** Code(s)</th>
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<tbody>
<tr>
<td>MEPI1</td>
<td>Epilepsy and Seizure Disorders: Sequencing Panel</td>
<td>81404 (x1), 81405 (x1), 81406 (x1)</td>
</tr>
<tr>
<td>DEPI1</td>
<td>Epilepsy and Seizure Disorders: Deletion/Duplication Panel</td>
<td>81304 (x1), 81403 (x1), 81404 (x1), 81405 (x1), 81406 (x1)</td>
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<tr>
<td>CMSNP</td>
<td>Chromosomal Microarray, CytoScan SNP Array</td>
<td>81229 (x1)</td>
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*Genes included on panels are subject to change. Some genes on this panel are associated with additional phenotypes. All genes on the next generation sequencing panel may be ordered separately.*

**Why Choose EGL Genetics?**
- The Epilepsy and Other Seizure Disorders Panel is the most comprehensive genetic test for epilepsy available.
- This panel was developed in collaboration with epilepsy genetic specialists.
- Free parental testing for up to two changes identified on the panel.
- Guaranteed 100% coverage with Sanger sequencing fill-in for low coverage regions.

**Test Code** | **Test Name**                                                                 | **CPT** Code(s)                          |
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**CPT** is a registered trademark of the American Medical Association.