Childhood Absence Epilepsy: \textit{GABRB3} Gene Deletion/Duplication

**Test Code:** DGABR  
**Turnaround time:** 2 weeks  
**CPT Codes:** 81228 x1

### Condition Description

Childhood absence epilepsy (CAE), which is a common idiopathic generalized epilepsy, accounts for 10-20\% of all epilepsy in children under the age of 16 years. Absence seizures are characterized by a brief loss of consciousness lasting between three and ten seconds and can occur up to 200 times a day. Tanaka et al. (2008) identified three missense mutations in the \textit{GABRB3} gene (15q11.2-q12) in four out of 48 families (8\%) with remitting CAE.

For patients with suspected childhood absence epilepsy, sequence analysis is recommended as the first step in mutation identification. For patients in whom mutations are not identified by full gene sequencing, deletion/duplication analysis is appropriate.

**References:**
- OMIM \#137192: \textit{GABRB3} gene
- OMIM \#612269: CAE

### Genes

\textit{GABRB3}

### Indications

This test is indicated for:
- Confirmation of a clinical diagnosis of childhood absence epilepsy in an individual in whom sequence analysis was negative.
- Carrier testing in adults with a family history of childhood absence epilepsy in whom sequence analysis was negative.

### Methodology

DNA isolated from peripheral blood is hybridized to a CGH array to detect deletions and duplications. The targeted CGH array has overlapping probes which cover the entire genomic region.

Please note that a "backbone" of probes across the entire genome are included on the array for analytical and quality control purposes. Rarely, off-target copy number variants causative of disease may be identified that may or may not be related to the patient's phenotype. Only known pathogenic off-target copy number variants will be reported. Off-target copy number variants of unknown clinical significance will not be reported.

### Detection

Detection is limited to duplications and deletions. The CGH array will not detect point or intronic mutations. Results of molecular analysis must be interpreted in the context of the patient's clinical and/or biochemical phenotype.

### Specimen Requirements

Submit only 1 of the following specimen types

* Preferred specimen type: Whole Blood

**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: Refrigerate until time of shipment. Ship sample within 5 days of collection at room temperature with overnight delivery.

**Type: Saliva**

Specimen Requirements:
Oragene™ Saliva Collection kit (available through EGL) used according to manufacturer instructions.

Specimen Collection and Shipping: Store sample at room temperature. Ship sample within 5 days of collection at room temperature with overnight delivery.

**Special Instructions**

Sequence analysis is required before deletion/duplication analysis by targeted CGH array. If sequencing is performed outside of EGL Genetics, please submit a copy of the sequencing report with the test requisition.

**Related Tests**

- Sequence analysis of the GABRB3 gene is available and is required before deletion/duplication analysis.
- 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.