X-linked Epilepsy with Variable Learning Disabilities and Behavior Disorders: SYN1 Gene Deletion/Duplication

Test Code: DSYN1
Turnaround time: 2 weeks
CPT Codes: 81228 x1

Condition Description

A mutation in the SYN1 gene (Xp11.4-p11.2) has been described in members of a family with epilepsy, variable learning disabilities, and behavior disorders. Some of the affected males were of normal intelligence with epilepsy only and others had various combinations of epilepsy, learning difficulties, macrocephaly, and aggressive behavior. The natural history of the epilepsy was variable. Some individuals had episodes only during childhood, others developed epilepsy later in life, and others had epilepsy only in association with specific stimuli.

The SYN1 gene encodes the synapsin I protein, a synaptic vesicle associated protein involved in the regulation of neurotransmitter release.

For patients with suspected X-linked epilepsy with variable learning disabilities and behavior disorders, 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 #300497: Epilepsy, X-Linked, with variable learning disabilities and behavior disorders.
- OMIM #313440: Synapsin I.

Genes

SYN1

Indications

This test is indicated for:
- Confirmation of a clinical diagnosis of X-linked epilepsy with variable learning disabilities and behavior disorders in an individual in whom sequence analysis was negative.
- Carrier testing in adults with a family history of X-linked epilepsy with variable learning disabilities and behavior disorders 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) or ACD (yellow 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

Submit copies of diagnostic biochemical test results with the sample, if appropriate. Contact the laboratory if further information is needed.

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

Related Tests

- Sequence analysis of the SYN1 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.