XLMR 91: ZDHHC15 Gene Deletion/Duplication

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

Condition Description

A female with a translocation resulting in the loss of ZDHHC15 expression has been described in the literature. This individual had severe muscular hypotonia in infancy, early childhood obesity, small hands and feet, facial changes and delayed psychomotor development. As an adult, she was reported to be of average height and weight but had severe psychomotor delays. Additionally, she had seizures, dysmorphic facies, and no speech. The t(X;15)(q13.3;cen) balanced translocation in this individual resulted in the ZDHHC15 transcript being absent from lymphocytes. Methylation studies revealed 100% skewed X inactivation with the normal X chromosome inactive.

For patients with suspected XLMR 91, 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:

Genes

ZDHHC15

Indications

This test is indicated for:

- Confirmation of a clinical diagnosis of XLMR 91 in an individual in whom sequence analysis was negative.
- Carrier testing in adults with a family history of XLMR 91 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 EGL Genetics, please submit a copy of the sequencing report with the test requisition.

Related Tests

- Sequence analysis of the ZDHHC15 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.