RPL10-Associated Autism Spectrum Disorder: RPL10 Gene Deletion/Duplication

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

Condition Description

The RPL10 gene (Xq28, previously known as the QM gene) encodes ribosomal protein L10, a highly conserved component of the large subunit of the ribosome that plays an important role in protein synthesis. It is located at Xq28, which is within a candidate region for autism spectrum disorders (ASD). Klauck et al. (2006) reported that different missense mutations in the RPL10 gene were identified in two sets of brothers with ASD from two unrelated families. Both of these mutations were located in exon 7 at the C-terminal end of the gene.

For patients with suspected RPL10-Associated ASD, 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.

Reference:

Genes

RPL10

Indications

This test is indicated for:
- Confirmation of a clinical diagnosis of RPL10-Associated ASD in an individual in whom sequence analysis was negative.
- Carrier testing in adults with a family history of RPL10-Associated ASD 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.

Related Tests

- Sequence analysis of the \textit{RPL10} 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.
- X-Linked Intellectual Disability panels are available for 30, 60, and 90 genes.