**Condition Description**

Both deletions and point mutations in the *IL1RAPL1* gene (Xp22.1-p21.3) have been associated with X-linked mental retardation (XLMR). Affected males usually have mild to severe nonsyndromic XLMR without other abnormalities, dysmorphic features, or neurological findings. Hyperactivity and self-aggressive behavior have been reported. Females in some families have been reported to have MR, while females in other families appear to be unaffected.

*IL1RAPL1* may also be deleted in families with a contiguous gene deletion syndrome that includes MR, adrenal hypoplasia, Duchenne muscular dystrophy, and glycerol kinase deficiency.

For patients with suspected XLMR 21, 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**

*IL1RAPL1*

**Indications**

This test is indicated for:

- Confirmation of a clinical diagnosis of XLMR 21 in individuals who have tested negative for sequence analysis
- Carrier testing in adult females with a family history of XLMR 21 who have tested negative for sequence analysis

**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**

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 *IL1RAPL1* gene is available.
- Prenatal testing is available to couples who are confirmed carriers of mutations. Please contact the laboratory genetic counselor to discuss appropriate testing prior to collecting a prenatal specimen.