XLMR, KDM5C-related: KDM5C Gene Deletion/Duplication

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

**Condition Description**

Mutations in the KDM5C gene (Xp11.22-p11.21) have been shown to cause an X-linked recessive syndromic mental retardation syndrome. Phenotypic features that have been reported include facial hypotonia, maxillary hypoplasia, strabismus, large ears with raised lobes, big hands with large fingers and proximal thumbs, prominent and separated superior incisors, scrotal tongue, and pectus excavatum. Other features of this syndrome include slowly progressive spastic paraplegia, epileptic seizures, short stature, microcephaly, hypermetropia, and small feet, testes, and penis. Aggressive behavior and an overfriendly and anxious character have also been reported.

The phenotype associated with mutations in the KDM5C gene is variable with regard to dysmorphism and cognitive impairment. In some families, the X-linked mental retardation seems to be nonsyndromic, with no dysmorphic features. It has been estimated that the frequency of mutations in the KDM5C gene may account for 2.8% to 3.3% of families with XLMR.

[Click here](#) for the OMIM summary on this condition.

### Genes

**KDM5C**

### Indications

This test is indicated for:

- Confirmation of a clinical diagnosis of KDM5C-related syndromic XLMR in individuals who have tested negative for sequence analysis
- Carrier testing in adult females with a family history of KDM5C-related syndromic XLMR 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.

### 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**

**Type: Whole Blood (EDTA)**

**Specimen Requirements:**
- EDTA (Purple Top)  
- Infants and Young Children (2 years of age to 10 years old): 3-5 ml  
- Older Children & Adults: 5-10 ml  
- Autopsy: 2-3 ml uncotted cord or cardiac blood

**Specimen Collection and Shipping:**
Ship sample at room temperature for receipt at EGL within 24 hours of collection. Do not refrigerate or freeze.

**Type: DNA, Isolated**

**Specimen Requirements:**
- Microtainer 3µg  
- Isolation using the Perkin Elmer™Chemagen™ Chemagen™ Automated Extraction method or Qiagen™ Puregene kit for DNA extraction is recommended.

**Specimen Collection and Shipping:**
Refrigerate until time of shipment in 100 ng/µL in TE buffer. Ship sample 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

- Sequencing analysis of the *KDM5C* gene is available (YJ) and is required before deletion/duplication analysis.
- ACGH array-based test for deletion/duplication analysis of 64 different X-linked intellectual disability genes is available (OL).
- Prenatal testing is available to adult females who are confirmed carriers of mutations. Please contact the laboratory genetic counselor to discuss appropriate testing prior to collecting a prenatal specimen.