XLMR with Growth Hormone Deficiency: SOX3 Gene Deletion/Duplication

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

**Condition Description**

Duplications of and in the SOX3 gene (Xq26.3) have been associated with X-linked recessive mental retardation with growth hormone deficiency. Mental retardation is mild to moderate and growth hormone levels can vary between affected individuals even within families. Some affected males have died during the first day of life and exhibited postmortem findings of hypopituitarism, presumably due to hypopituitarism. Others have variable combinations of hypothyroidism, delayed pubertal development, and short stature due to growth hormone deficiency. All surviving affected individuals exhibited mild to moderate mental retardation. Behavior in affected individuals has been considered infantile.

Variable craniofacial dysmorphism has also been reported, including hypertelorism, epicanthus, synophrys, broad nasal bridge, high-arched palate, long philtrum, cup helices, and coarse facies. In one study, male patients continued to grow until well into their twenties and reached a height ranging from 135 to 159 cm. Brain MRI in some affected males showed anterior pituitary hypoplasia, ectopic posterior pituitary, and absent infundibulum.

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

**Genes**

SOX3

**Indications**

This test is indicated for:

- Confirmation of a clinical/biochemical diagnosis of XLMR with growth hormone deficiency in individuals who have tested negative for sequence analysis
- Carrier testing in adult females with a family history of XLMR with growth hormone deficiency 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**

- Sequencing analysis of the SOX3 gene is available (YV) and is required before deletion/duplication analysis.
- X-Linked Intellectual Disability panels are available for 30, 60, and 90+ genes.
- 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.