**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 hypoadrenalism, 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.

**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 unclotted cord or cardiac blood

**Specimen Collection and Shipping:**

Ship sample at room temperature for receipt at EGL within 72 hours of collection. Do not 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 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.