BCOR-related Disorders: BCOR Gene Deletion/Duplication

**Condition Description**

**Lenz microphthalmia syndrome**

Lenz microphthalmia syndrome (LMS) is an X-linked disorder characterized by unilateral or bilateral microphthalmia and/or anophthalmia. Extraocular malformations may affect the ears, teeth, fingers, skeleton, and genitourinary system. Other features can include glaucoma, coloboma, hearing loss, microcephaly, webbed neck, and cleft lip/palate. Developmental delay or intellectual disability is present in approximately 60% of affected males. BCOR is the only gene known to be associated with LMS although another X-linked locus is also known to be associated. Female carriers are typically not affected.

**Oculofaciocardiodental syndrome**

Oculofaciocardiodental (OFCD) syndrome (OMIM #300166) is also associated with mutations in BCOR. OFCD syndrome affects females and is lethal in males. Characteristics include congenital cataracts with or without unilateral/bilateral microphthalmia, long narrow face, cleft palate, cardiac defects, and dental anomalies. Microphthalmia is less severe in OFCD syndrome than in LMS. Females with OFCD syndrome often have normal intelligence. Known BCOR mutations causing OFCD include point mutations, small deletions, and deletions involving one or more exons.

For patients with suspected BCOR-related disorders, 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:**

- GeneReviews
- OMIM #300166: OFCD syndrome
- OMIM #300485: BCOR gene

**Genes**

**BCOR**

**Indications**

This test is indicated for:

- Confirmation of a clinical diagnosis of a BCOR-related disorder in an individual in whom sequence analysis was negative.
- Carrier testing in adults with a family history of a BCOR-related disorder 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.

**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: 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.

**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.

**Related Tests**

- Sequence analysis of the BCOR 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.