Anophthalmia/Microphthalmia/Anterior Segment Dysgenesis/Anomaly: Deletion/Duplication Panel

Test Code: MD139  
Turnaround time: 2 weeks  
CPT Codes: 81228 x1

**Condition Description**

Anophthalmia generally refers to an absence of the globe while the eyelids, conjunctiva, and lacrimal glands remain. Microphthalmia is a heterogeneous group of malformations with reduction in the size of the eyeball that is anatomically intact with only axial length reduction (simple form), or can also include anterior segment dysgenesis (complex form). Both anophthalmia and microphthalmia can occur as isolated or syndromic and can be bilateral or unilateral. Anterior segment dysgenesis generally refers to a complex spectrum of anomalies such as Axenfeld-Rieger anomaly and Peters anomaly where axial length may not be severely compromised. Syndromic forms can include Fraser syndrome, microphthalmia with linear skin defects, and Manitoba oculotrichoanal syndrome (MOTA).

**References:**

- OMIM
- GeneReviews

**Genes**

B3GALT5, BCOR, BMP4, COL4A1, CYP1B1, FOXC1, FOXE3, FRAS1, FREM1, FREM2, GRIP1, HCCS, MFRP, NDP, OTX2, PAX6, PITX2, PITX3, SMOC1, SOX2, STRA6, VAX1, VSX2

**Indications**

This test is indicated for:

- Confirmation of a clinical diagnosis of anophthalmia, microphthalmia, or anterior segment dysgenesis/anomaly.
- Carrier testing in adults with a family history of anophthalmia, microphthalmia, or anterior segment dysgenesis/anomaly.

**Methodology**

**Deletion/Duplication Analysis:** 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**

**Deletion/Duplication Analysis:** 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.

**Special Instructions**

Please include fundus photographs, electroretinogram (ERG) findings, visual field findings, and visual acuity, if available, for expert review and clinical
correlation with test results.

**Related Tests**

- Anophthalmia/Microphthalmia/Anterior Segment Dysgenesis/Anomaly: Sequencing Panel
- Eye Disorders: Comprehensive Sequencing
- Eye Disorders: Deletion/Duplication Panel