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

B3GLCT, 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. 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**

**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: 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: Ship sample at room temperature with overnight delivery.

#### Type: Isolated DNA

Specimen Requirements:
- In microtainer: 10 ug

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Isolation using the Qiagen™ Puregene kit for DNA extraction is recommended.

Specimen Collection and Shipping: Refrigerate until time of shipment in 100 ng/ul of TE buffer. Ship sample at room temperature with overnight delivery.

### 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