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**Lab Dept:**                    **Anatomic Pathology**

**Test Name:**                **ANOPHTHALMIA, MICROPHTHALMIA (SOX2)  
DELETION/DUPLICATION**

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***General Information***

**Lab Order Codes:**        SOCD (Blood samples only)

**Synonyms:**                Developmental Eye Disorders Gene Testing

**CPT Codes:**                81479 – Molecular Pathology, unlisted

**Test Includes:**            For the gene of interest, the number of copies present in the patient's sample.

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***Logistics***

**Test Indications:**        Several developmental eye disorders have a known genetic basis including microphthalmia and anophthalmia.

**Applications gene copy number analysis:**

1. As a complementary test or substitute for FISH and quantitative microarray analysis when a deletion or duplication syndrome (contiguous or single gene) is suspected.
2. As a confirmation test for a genomic gain or loss identified by genome-wide microarray analysis.
3. To determine the presence or absence of a specific gene within a deleted chromosomal segment (contiguous gene deletion).
4. Diagnostic testing in a Mendelian disorder due to functional loss of one allele (haploinsufficiency), in particular when sequence analysis failed to identify a causative mutation and a whole gene deletion is a possible cause.
5. Carrier testing for deletion/duplication syndromes when the specific region is known in an affected relative.

**Lab Testing Sections:**    Anatomic Pathology - Sendouts

**Referred to:**                GeneDx, Inc. (GDx Test: 906, Specify gene/mutation)

**Phone Numbers:**        MIN Lab: 612-813-6280

STP Lab: 651-220-6550

**Test Availability:**        Daily, 24 hours. Specimens collected Saturday or Sunday will be held for shipment on Monday.

**Turnaround Time:**        3 - 4 weeks

**Special Instructions:** A GeneDx signed [request form](#) must be sent with any patient or specimen to the laboratory.

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### ***Specimen***

**Specimen Type:** Whole blood

**Container:** Lavender top (EDTA) tube

**Draw Volume:** 1 - 5 mL blood

**Processed Volume:** Same as Draw Volume

**Collection:** Routine blood collection, invert gently to mix

**Special Processing:** Lab Staff: Send whole blood in original collection container labeled with patient name, date of birth and medical record number, including signed consent form and requisition, with a cool pack in warm weather, via overnight or second-day courier so that the sample will arrive at GeneDx, Monday through Saturday. Samples drawn on Saturday or Sunday should be held at refrigerated temperatures for shipment on Sunday. **Do not** freeze.

**Note:** Specimens may be stored at refrigerated temperatures for up to 7 days prior to shipping.

**Patient Preparation:** None

**Sample Rejection:** Unrefrigerated specimens older than 48 hours; clotted or hemolyzed for blood; frozen specimens; mislabeled or unlabeled specimens

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### ***Interpretive***

**Reference Range:** Interpretive report

**Critical Values:** N/A

**Limitations:** N/A

**Methodology:** Comparative quantitative analysis, fluorescent quantitative PCR-based method. Analysis done in triplicate.

**References:** [GeneDx, Inc.](#) January 2018  
(301) 519-2100 Fax (301) 519-2892

**Updates:** 1/29/2013: CPT 2013 update  
3/26/2014: CPT update