ANOPHTHALMIA, MICROPHTHALMIA (SOX2) DELETION/DUPLICATION

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.

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 genomewide 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.

**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

**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