
Lab Dept: **Anatomic Pathology**

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

General Information

Lab Order Codes: SOCD (Blood samples only)

Synonyms: Developmental Eye Disorders Gene Testing

CPT Codes: 81400 – Molecular Pathology, Level 1

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 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 (Preferred draws are Sunday - Thursday as specimens can only be received at the reference lab Monday - Friday. Specimens collected Friday or Saturday will be held for shipment on Monday.)

Turnaround Time: 4 - 6 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 venipuncture for blood specimens, 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, Inc. on a weekday (Monday through Friday). Samples drawn on Friday or Saturday 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: Buccal specimens are not acceptable for this testing.

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

References: [GeneDx, Inc.](#) March 2012
(301) 519-2100 Fax (301) 519-2892

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