



Laboratory Service Manual

Lab Dept: Anatomic Pathology

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

General Information

Lab Order Codes: SOCD (Blood samples only)

Synonyms: Developmental Eye Disorders Gene Testing

CPT Codes: 83891 x2 – Isolation or extraction of highly purified nucleic acid
83892 x2 – Molecular diagnostics; enzymatic digestion
83898 x12 – Amplification of nucleic acid

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 of CopyDx 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#: 904, Specify gene/mutation)

Phone Numbers:

Minneapolis: 612-813-6280

Saint Paul: 651-220-6550



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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 consent form and request form must be sent with any patient or specimen to the laboratory.

Specimen

Specimen Type:	Whole blood
Container:	Blood: 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, 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 3 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. February 2008 (301) 519-2100 Fax (301) 519-2892



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