
Lab Dept: **Anatomic Pathology**

Test Name: **ANOPHTHALMIA, MICROPHTHALMIA (SIX6)
DELETION/DUPLICATION**

General Information

Lab Order Codes: SICD (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.
1. Confirmation of clinical diagnosis
2. Genetic counseling
3. Prenatal diagnosis

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, via overnight or second-day courier so that the sample will arrive at GeneDx, Inc. on a weekday (Monday through Friday). Include a cool pack when shipping in warm weather. 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:	Buccal brushes are not acceptable for this testing.
Methodology:	Targeted array CGH analysis with exon-level resolution
References:	GeneDx, Inc. February 2012 (301) 519-2100 Fax (301) 519-2892
Updates:	1/29/2013: CPT 2013 Updates 3/26/2014: CPT 2014 Updates