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<th>Lab Dept:</th>
<th>Anatomic Pathology</th>
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<td>Test Name:</td>
<td>ANOPHTHALMIA, MICROPHTHALMIA (SOX2) SEQUENCING</td>
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**General Information**

**Lab Order Codes:**
SOX (Blood or Buccal Swab)

**Synonyms:**
Developmental Eye Disorders

**CPT Codes:**
81479 – Unlisted Molecular Pathology procedure (SOX2 Sequencing)

**Test Includes:**
Using genomic DNA, bi-directional sequence of the coding portion of the SOX2 gene (single large exon) is obtained and analyzed. Mutation found in the first of a family to be tested and is confirmed by repeat analysis using sequencing, restriction fragment analysis, or another appropriate method.

**Logistics**

**Test Indications:**
Several developmental eye disorders have a known genetic basis including microphthalmia and anophthalmia. Anophthalmia is the complete absence of the globe, or bulb, of the eye and hence the most structural eye malformation. A milder form is microphthalmia, where total axial length of the eye globe is the least two standard deviations below the mean for age. Simple microphthalmia refers to a structurally normal eye with short total axial length. In each of these conditions, the eyelids, conjunctiva and lacrimal apparatus are normal. In complex microphthalmia, additional abnormalities are present and may include anterior segment dysgenesis, cataract, persistent hyperplastic primary vitreous, chorioretinal coloboma and/or retinal dysplasia. In addition, anophthalmia and microphthalmia may be seen in association with various genetic syndromes or chromosome abnormalities. Mutations in the SOX2 and SIX6 genes leading to haploinsufficiency may be associated with hearing loss, developmental delay, esophageal atresia, genitourinary abnormalities, myopathy, and spastic diplegia. Hemizygosity for SIX6 has been seen in some cases of bilateral anophthalmia due to interstitial chromosome deletions. Homozygosity for PAX6 mutation has also been associated with anophthalmia.

**Reasons for referral:**
1. Confirmation of a clinical diagnosis
2. Genetic counseling

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

**Referred to:**
GeneDx, Inc. (GDX: 132)
**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 weeks

**Special Instructions:**

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

SOX2 can be tested using cells obtained by swabbing the buccal mucosa (inside of cheek). Buccal specimens are not accepted on children under 6 months of age. Buccal swab collection kits are available from GeneDx (ph. 301-519-2100).

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### Specimen

**Specimen Type:** Whole blood (preferred specimen)

Buccal cell swab

**Container:**

Blood: Lavender top (EDTA) tube

Buccal Smear: Cytobrush Plus Cell Collector® kit

**Draw Volume:**

1 - 5 mL blood

2 swabs from the Cytobrush Plus Cell Collector® kit

**Processed Volume:**

Same as Draw Volume

**Collection:**

Routine blood collection for blood specimens, invert gently to mix

Buccal swab kit for buccal swabs:

**Buccal Cell Collection procedure:**

1. Remove a swab from the Buccal swab kit touching only the “stick” end.
2. **Do Not** rinse mouth before starting. Have the individual open his/her mouth. Twirl the swab on the inner cheek for 30 seconds. **Do Not** scrape so hard that the cheek bleeds.
3. Place the swab end in the labeled tube. Cut the “stick” with scissors at the level of the top of the tube. Replace cap and close completely.
4. Repeat the process with another swab on the opposite cheek.
5. Send the kit to the lab for processing and mailing.
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 Monday. **Do not** freeze.

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

Mail the Cytobrush Plus Cell Collector® kit, including signed consent form and requisition, by regular mail to GeneDx in the included envelope.

Patient Preparation: For buccal cell collection, **Do Not** have the patient rinse his/her mouth.

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

**Interpretive**

Reference Range: No mutations detected

Critical Values: N/A

Limitations: Buccal specimens are not acceptable on children under 6 months of age.

Methodology: Capillary sequencing

References: [Gene Dx, Inc](http://www.genedx.com) January 2018
(301) 519-2100  Fax (301) 519-2892

Updates: 1/29/2013: CPT 2013 update