



Laboratory Service Manual

Lab Dept: Anatomic Pathology

Test Name: ANIRIDIA (PAX6) GENE SEQUENCING

General Information

Lab Order Codes: PAX (Blood or Buccal Swab)

Synonyms: Developmental Eye Disorders Gene Testing

CPT Codes: 83891 x16 – Isolation or extraction of highly purified nucleic acid

83894 x32 – Separation by gel electrophoresis

83892 x3 – Molecular diagnostics; enzymatic digestion

83898 x32 – Amplification of nucleic acid

83904 x32 – Mutation identification by sequencing, single segment

Test Includes: Using genomic DNA, bi-directional sequence of the coding portion of the PAX6 gene (exons 4-14) and splice junctions 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. If negative, gene copy analysis (PAX6 CopyDx) is available for identifying submicroscopic whole or partial PAX6 gene deletions.

Logistics

Test Indications: Aniridia is a developmental anomaly of the entire eye, characterized by varying degrees of iris hypoplasia, foveal hypoplasia, and/or nysagmus. Frequently associated findings with onset later in life include cataracts, limbal stem cell deficiency, and glaucoma. The most severe presentation of aniridia is the complete absence of the iris. Milder disease may include enlargement and irregularity of the pupil and small, slit-like defects in the anterior layer. Vision is variably affected, and the severity of vision loss tends to correlate with the presence of other associated defects.

Reasons for referral:

1. Confirmation of clinical diagnosis
2. Determination of the molecular basis of aniridia in patients at risk for Wilm's tumor
3. Genetic counseling

Lab Testing Sections: Anatomic Pathology - Sendouts

Referred to: GeneDx, Inc. (GDX#: 1311)



Laboratory Service Manual

Phone Numbers:

Minneapolis: 612-813-6280

Saint Paul: 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: 8-10 weeks for new patients

Special Instructions: A signed consent form must be sent with any specimen. Forms can be obtained through the following link:
http://www.genedx.com/required_forms.php

PAX6 can be tested using cells obtained by swabbing the buccal mucosa (inside of cheek). Buccal swab collection kits are available from GeneDx (ph. 301-519-2100). Buccal Kit info link:
http://www.genedx.com/buccal_swab_kits.php

Specimen

Specimen Type: Whole blood

Buccal cell swab (preferred specimen)

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 venipuncture 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.



Laboratory Service Manual

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, 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.

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:

N/A

Methodology:

Bi-directional sequence analysis

Contraindications:

N/A

References:

GeneDx, Inc. Web Page www.genedx.com February 2008