



# Laboratory Service Manual

**Lab Dept:** Anatomic Pathology

**Test Name:** ANIRIDIA (PAX6) COPY DX  
DELETION/DUPLICATION

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## **General Information**

**Lab Order Codes:** PXCD (Blood samples only)

**Synonyms:** Aniridia Gene Testing; 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.

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## **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.

### **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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<b>Test Availability:</b>	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.)
<b>Turnaround Time:</b>	4 - 6 weeks
<b>Special Instructions:</b>	A GeneDx signed <a href="#">consent form</a> and <a href="#">request form</a> must be sent with any patient or specimen to the laboratory.

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## ***Specimen***

<b>Specimen Type:</b>	Whole blood
<b>Container:</b>	Blood: Lavender top (EDTA) tube
<b>Draw Volume:</b>	1 - 5 mL blood
<b>Processed Volume:</b>	Same as Draw Volume
<b>Collection:</b>	Routine venipuncture for blood specimens, invert gently to mix
<b>Special Processing:</b>	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. <b>Do not</b> freeze. <b>Note:</b> Specimens may be stored at refrigerated temperatures for up to 3 days prior to shipping.
<b>Patient Preparation:</b>	None
<b>Sample Rejection:</b>	Unrefrigerated specimens older than 48 hours, clotted or hemolyzed for blood, frozen specimens, mislabeled or unlabeled specimens

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## ***Interpretive***

<b>Reference Range:</b>	Interpretive report
<b>Critical Values:</b>	N/A
<b>Limitations:</b>	N/A
<b>Methodology:</b>	Comparative quantitative analysis, fluorescent quantitative PCR-based method. Analysis done in triplicate.
<b>References:</b>	<a href="#">GeneDx, Inc.</a> February 2008 (301) 519-2100 Fax (301) 519-2892



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