
Lab Dept: **Anatomic Pathology**

Test Name: **POPLITEAL PTERYGIUM SYNDROME (IRF6 EXON 4 ONLY) SEQUENCING**

General Information

Lab Order Codes: IRF4 (Blood or Buccal Swab)

Synonyms: Popliteal pterygium syndrome; PPS; cleft lip/palate; paramedian mucous cysts of the lower lip; digital and genital abnormalities; faciogenitalpopliteal syndrome

CPT Codes: 81479 – Molecular Pathology Unlisted procedure

Test Includes: Analysis is performed by bi-directional sequencing of the coding regions and splice sites of exon 4 of the IRF6 gene for patients with a clinical diagnosis of PPS. Mutations found in the first person of a family to be tested are confirmed by repeat analysis using sequencing, restriction fragment analysis, or another appropriate method.

Logistics

Test Indications: As with Van der Woude syndrome, cleft lip, cleft palate and lip pits are included in the clinical spectrum of Popliteal Pterygium Syndrome. In addition to the orofacial features, 90% of individuals with PPS have popliteal web present and ~50% of patients have genital abnormalities. Toenail dysplasia, syndactyly of the toes and digits, and bony deformities also may be present.

Reasons for referral:

1. Confirmation of clinical diagnosis
2. Genetic counseling
3. Identification of a genetic basis for cleft lip/palate in the patient/family.

Lab Testing Sections: Anatomic Pathology - Sendouts

Referred to: GeneDx, Inc. (GDX#: 248)

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 weeks for new patients

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

IRF6 exon 4 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).

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.
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, Inc. on a weekday (Monday through Friday). Samples drawn on Friday or Saturday should be held at refrigerated temperatures for shipment on Sunday. Refrigerated specimens can be held for 7 days. **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: Buccal specimens can only be used for sequencing, they are not appropriate for deletion/duplication testing.

Methodology: Bi-directional sequence analysis

References: [GeneDx, Inc.](#) February 2012
(301) 519-2100 Fax (301) 519-2892

Updates: 2/7/2013: CPT update