



**MAYO**  
Mayo Medical Laboratories  
1-800-533-1710

**NEW TEST ANNOUNCEMENT**  
**NOTIFICATION DATE:** February 19, 2014  
**EFFECTIVE DATE:** February 25, 2014

## **FLG Gene, Mutation Analysis**

Test ID: FLGA

**USEFUL FOR:** Genetic diagnosis of ichthyosis vulgaris for clinical management, risk assessment for atopic diseases and atopic disease-associated asthma, and genetic counseling for family members

**METHODOLOGY:** Polymerase Chain Reaction (PCR) Followed by DNA Sequence Analysis

**REFERENCE VALUES:** An interpretive report will be provided.

**SPECIMEN REQUIREMENTS:** Specimen preferred to arrive within 96 hours of collection.

**Submit only 1 of the following specimens:**

**Specimen Type:** Blood

**Container/Tube:**

**Preferred:** Lavender top (EDTA) or yellow top (ACD)

**Acceptable:** Any anticoagulant

**Specimen Volume:** 3 mL

**Collection Instructions:**

1. Invert several times to mix blood.
2. Send specimen in original tube.

**Specimen Stability Information:** Ambient (preferred)/Refrigerated/Frozen

**Specimen Type:** Cultured fibroblasts

**Container/Tube:** T-75 or T-25 flask

**Specimen Volume:** 1 Full T-75 or 2 full T-25 flasks

**Specimen Stability Information:** Ambient (preferred)/Refrigerated <24 hours

**Specimen Type:** Skin biopsy

**Container/Tube:** Sterile container with any standard cell culture media (eg, minimal essential media, RPMI 1640). The solution should be supplemented with 1% penicillin and streptomycin. Tubes can be supplied upon request (Eagle's minimum essential medium with 1% penicillin and streptomycin [Supply T115]).

**Specimen Volume:** 4-mm punch

**Specimen Stability Information:** Refrigerated (preferred)/Ambient

**NOTE:**

- New York Clients-Informed consent is required. Please document on the request form or electronic order that a copy is on file. An Informed Consent for Genetic Testing (Supply T576) is available in Special Instructions.
- Molecular Genetics-Congenital Inherited Diseases Patient Information Sheet (Supply T521) in Special Instructions

**SPECIMEN STABILITY INFORMATION:**

Specimen Type	Temperature	Time
Varies	Varies	

**CAUTIONS:**

- This assay does not detect all of the mutations in filaggrin (FLG) gene or any other gene mutations that cause ichthyosis vulgaris. Therefore, the absence of a detectable mutation does not rule out the possibility that an individual is a carrier of, or affected with, ichthyosis vulgaris. For carrier testing, it is important to first document the presence of an FLG gene mutation in an affected family member.
- Test results should be interpreted in context of clinical findings, family history, and other laboratory data. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.
- Rare polymorphisms exist that could lead to false-negative or positive results. If results obtained do not match the clinical findings, additional testing should be considered.
- Bone marrow transplants from allogenic donors will interfere with testing. Call Mayo Medical Laboratories for instructions for testing patients who have received a bone marrow transplant.

**CPT CODE:** 81401-FLG (filaggrin) (eg, ichthyosis vulgaris), common variants (eg, R501X, 2282del4, R2447X, S3247X, 3702delG)

**DAY(S) SET UP:** Monday; 10 a.m.

**ANALYTIC TIME:** 10 days

**NOTE:** The following referral test code(s) will become obsolete effective March 25, 2014.

Test Name	Test ID	Referral Lab Code	Referral Lab
FLG Gene Analysis in Ichthyosis Vulgaris with or without Atopic Dermatitis	ZW168	123	GeneDx, Inc.

**QUESTIONS:** Contact your Mayo Medical Laboratories' Regional Manager or Marvin H. Anderson, Jr., MML Laboratory Technologist Resource Coordinator  
Telephone: 800-533-1710