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**Reporting Title: FLG Gene, Mutation Analysis**  
**Performing Location: Rochester****Specimen Requirements:**

Forms:

1. 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.
2. Molecular Genetics-Congenital Inherited Diseases Patient Information Sheet (Supply T521) in Special Instructions

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 &lt;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

Specimen Type	Temperature	Time
Varies	Varies	

**Ask at Order Entry (AOE) Questions:**

Test ID	Question ID	Description	Type	Reportable
FBC	CG030	Source: <ul style="list-style-type: none"> <li>• Autopsy</li> <li>• Chorionic Villi</li> <li>• Products of Conception or Stillbirth</li> </ul>	Answer List	No
FBC	CG033	Reason for Referral	Plain Text	No

**Result Codes:**

Result ID	Reporting Name	Type	Unit	LOINC®
55013	Reason for Referral	Alphanumeric		In Process
55014	Result	Alphanumeric		In Process
55015	Interpretation	Alphanumeric		In Process
55016	Method	Alphanumeric		In Process
55017	Amendment	Alphanumeric		In Process
55018	Specimen	Alphanumeric		In Process
55019	Source	Alphanumeric		In Process
55020	Reviewed By	Alphanumeric		In Process
55021	Release Date	Alphanumeric		In Process

**CPT Code Information:**

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

**Reflex Tests:**

Test ID	Reporting Name	CPT Units	CPT Code	Always Performed	Orderable Separately
FBC	Fibroblast Culture for Genetic Test		Profile	No	Yes

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**Result Codes for Reflex Tests:**

Test ID	Result ID	Reporting Name	Type	Unit	LOINC®
FBC	16167	Specimen	Alphanumeric		N/A
FBC	16337	Specimen ID	Alphanumeric		N/A
FBC	CG030	Source	Alphanumeric		31208-2
FBC	16168	Order Date	Alphanumeric		N/A
FBC	CG033	Reason For Referral	Alphanumeric		42349-1
FBC	16172	Method	Alphanumeric		49549-9
FBC	16175	Interpretation	Alphanumeric		69965-2
FBC	16176	Amendment	Alphanumeric		In Process
FBC	16177	Consultant	Alphanumeric		N/A
FBC	16178	Report Date	Alphanumeric		N/A

**Reference Values:**

An interpretive report will be provided.