

Laboratory Service Report

1-800-533-1710

Patient Name SAMPLEREPORT,FLGA	Patient ID SA00065089	Age 57	Gender M	Order # SA00065089
Ordering Phys CLIENT,CLIENT				DOB 12/21/1956
Client Order # SA00065089	Account Information			Report Notes
Collected 01/13/2014 00:00	C7028846-DLMP Rochester SDSC 2 - Client Support			
Printed 02/19/2014 15:54	Rochester, MN 55901			

Test Flag Results Unit Value Site*

FLG Gene, Mutation Analysis

RECEIVED: 01/13/2014 12:19 **REPORTED:** 01/14/2014 13:21

Reason for Referral

MCR

Patient reported to have features suggestive of ichthyosis vulgaris or atopic dermatitis. Test for the presence of mutations in the FLG gene.

Result

MCR

The following heterozygous sequence change was detected:

Exon: 3

DNA change: c.1501C>T

Amino Acid change: p. R501X (Arg501STOP)

Classification: DELETERIOUS

Interpretation

MCR

This alteration is a known deleterious mutation.

This result indicates that this individual is a carrier of ichthyosis vulgaris (IV) and at an increased risk to develop atopic dermatitis (AD). This assumes that this individual is healthy and not clinically affected with these diseases.

Carriers of FLG mutations may exhibit none to very mild phenotype of IV. AD is a severe clinical phenotype that is observed in 37-50% of patients with two FLG alterations in trans (on opposite chromosomes). However, only approximately 8% of AD cases are associated with FLG alterations.

Since a mutation has been identified, testing of at risk family members is possible. If appropriate, genetic testing should be offered to this individual's reproductive partner to clarify their risk of having a child with disease.

A genetic consultation may be of benefit.

Unless reported or predicted to cause disease, alterations found deep in the intron or alterations that do not result in an amino acid substitution are not reported. These and common polymorphisms identified for this patient are available upon request.

CAUTIONS

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.

Performing Site Legend on Last Page of Report

Patient Name	Collection Date and Time	Report Status
SAMPLEREPORT,FLGA	01/13/2014 00:00	Final
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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.

Laboratory developed test.

Method

A PCR based assay was used to test for the following mutations in the FLG gene: c.1501C>T (R501X) and c.2282del4 (S761CfsX36).

Specimen Blood

Reviewed By

W Edward Highsmith Jr., PhD

Release Date 14 Jan 2014 13:19 MCR

* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director: Franklin R. Cockerill, III, M.D.

Patient Name	Collection Date and Time	Report Status
SAMPLEREPORT,FLGA	01/13/2014 00:00	Final
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