MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,FLGA	SA00065089	57	M	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 Rochester, MN 55901			
Printed 02/19/2014 15:54				

Test	Flag	Results	Unit	Reference Value	Perform 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.

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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).					MCR
Specimen		Blood			MCR
Reviewed By W Edward Highsmith Jr., PhD					MCR
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: