MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,FD	SA00007367	32	F	SA00007367
Ordering Phys				**DOB** 04/04/1980
Client Order # SA00007367	**Account Information**			**Report Notes**
Collected 07/31/2012	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 08/15/2012 14:09				

Test	Flag	Results	Unit	Reference Value	Perform Site*
Familial Dysautonomia, Mutation			REPORTED 08/14/2012 16:55		
Specimen		Blood			MCR
Specimen ID		976466			MCR
Order Date		01 Aug 2012 13:07			MCR
Reason For Referral					MCR
Carrier screen for familial dysautonomia. Test for the presence of a mutation in the IKBKAP gene.					
Method					MCR
A PCR-based assay was used to test for the following mutations in the IKBKAP gene: IVS20(+6)T>C and R696P.					
Results					MCR
One copy of the R696P mutation was identified.					
Interpretation					MCR
This result indicates that this individual is a carrier of familial dysautonomia. This interpretation assumes that this individual is healthy and is not clinically affected with familial dysautonomia.					
Since a mutation has been identified, genetic testing of at risk family members is possible. If appropriate, genetic testing should be offered to this individual's reproductive partner.					
A genetic consultation may be of benefit.					
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.					
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.					
Reviewed By:		D Brian Dawson PhD			MCR
Release Date		14 Aug 2012 16:54			MCR

Performing Site Legend on Last Page of Report

* Report times for Mayo performed tests are CST/CDT

TEST SITE

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>> Accession SA00007367 - Continued From Previous Page <<
>> Do Not Discard <<

* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director:

Patient Name	Collection Date and Time	Report Status
SAMPLEREPORT,FD	07/31/2012	Final
Page 2 of 2		** End of Report **

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TEST SITE