MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,SDHKM	SA00055264	46	F	SA00055264
Ordering Phys UNKNOWN,PROVIDER				**DOB** 06/10/1966
Client Order # SA00055264	**Account Information**			**Report Notes**
Collected 03/15/2013 01:11	C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901			
Printed 06/05/2013 08:00				

Test	Flag	Results	Unit	Reference Value	Perform Site*
SDH Known Mutation					
RECEIVED: 03/15/2013 01:11 **REPORTED:** 06/04/2013 18:01					
SDH Known Mutation					
Reason for Referral		Familial mutation			MCR
An SDHB gene mutation was previously identified in an affected family member. Test for the presence of the following mutation in the SDHB gene: Intron 6, c.642+1G>A (splice site mutation)					
Method		SDHB familial			MCR
Fluorescent DNA sequence analysis was used to test for the presence of a specific mutation in the SDHB gene, which was previously identified in an affected family member of this individual.					
Result		Positive			MCR
This individual was shown to be heterozygous for the familial mutation: Intron 6, c.642+1G>A (splice site mutation)					
Interpretation		SDHB positive			MCR
The above mutation was previously identified in an affected family member. The presence of this mutation in the individual tested here, therefore, is consistent with a diagnosis of hereditary paraganglioma-pheochromocytoma (PGL/PCC) syndrome for this individual. This individual is at risk for developing symptoms associated with PGL/PCC. Appropriate screening procedures should be considered. Since a mutation has been identified in the SDHB gene in this individual, genetic testing for this specific mutation in other family members is recommended. Please contact the Endocrine Laboratory at 1-800-533-1710 or the online test catalog at mayomedicallaboratories.com for information about how to order SDH Known Mutation (89554/SDHKM) for sequencing mutations.					
Comment		GC consult			MCR
A genetic consultation may be of benefit.					
Reviewed By		Dee Feighner			MCR
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. Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Misinterpretation of results may occur if the information provided is inaccurate or incomplete. A list of common polymorphisms identified for this patient is available from the Endocrine Laboratory upon request. Rarely, individuals may have a mutation or deletion in the gene(s) tested that is not identified by the described testing methodology. In addition, the phenotype observed in the individual tested here may be due to a variant in a gene not analyzed by this test. Laboratory developed test.					

Performing Site Legend on Last Page of Report

Patient Name	Collection Date and Time	Report Status
SAMPLEREPORT,SDHKM	03/15/2013 01:11	Final
		>> Continued on Next Page >>

* Report times for Mayo performed tests are CST/CDT

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SDH Known Mutation Sequencing		Performed			MCR

* 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,SDHKM	03/15/2013 01:11	Final
		** End of Report **

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