MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,SDHSP	SA00055293	46	F	SA00055293
Ordering Phys UNKNOWN,PROVIDER				**DOB** 06/10/1966
Client Order # SA00055293	**Account Information**			**Report Notes**
Collected 03/15/2013 01:22	C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901			
Printed 06/05/2013 08:12				

Test	Flag	Results	Unit	Reference Value	Perform Site*
SDH Gene Analysis					
RECEIVED: 03/15/2013 01:22 **REPORTED:** 06/04/2013 18:15					
SDH Gene Analysis					
Reason for Referral		Clinical features			MCR
Patient has clinical features suggestive of hereditary paraganglioma-pheochromocytoma (PGL/PCC) syndrome. Test for the presence of mutation(s) within the SDHB, SDHC, and SDHD genes.					
Method		Full gene sequencing			MCR
Fluorescent DNA sequence analysis was used to test for the presence of mutations in all exons and exon-intron boundaries of the SDHB, SDHC, and SDHD genes.					
Result		Positive mutation			MCR
This individual was shown to be heterozygous for the following mutation in the SDHD gene: Exon 2, c.112C>T, p.Arg38X (p.R38X)					
Interpretation		Positive			MCR
This result is consistent with a diagnosis of hereditary paraganglioma-pheochromocytoma (PGL/PCC) syndrome. Therefore, 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 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 or SDH Deletion Detection (89555/SDHDD) for large deletions of one or more exons.					
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.					
SDH Gene Sequencing		Performed			MCR

Performing Site Legend on Last Page of Report

Patient Name	Collection Date and Time	Report Status
SAMPLEREPORT,SDHSP	03/15/2013 01:22	Final
		>> Continued on Next Page >>

* Report times for Mayo performed tests are CST/CDT

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>> Accession SA00055293 - 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,SDHSP	03/15/2013 01:22	Final
Page 2 of 2		** End of Report **

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