MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,SDHSP	SA00055289	46	F	SA00055289
Ordering Phys UNKNOWN,PROVIDER				**DOB** 06/10/1966
Client Order # SA00055289	**Account Information**			**Report Notes**
Collected 03/15/2013 01:21	C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901			
Printed 06/05/2013 08:04				

Test	Flag	Results	Unit	Reference Value	Perform Site*
SDH Gene Analysis					
RECEIVED: 03/15/2013 01:21 **REPORTED:** 06/04/2013 18:13					
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		Sequencing and MLPA			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. Additionally, multiplex ligation dependent probe amplification (MLPA) was used to test for the presence of large deletions in the SDHB, SDHC, and SDHD genes. This method utilizes probes which cover all eight exons of the SDHB gene, all six exons of the SDHC gene, and all four exons of the SDHD gene.					
Result		Negative			MCR
No mutations or deletions were detected in the SDHB, SDHC, or SDHD genes.					
Interpretation		Negative			MCR
There is no genetic evidence for hereditary paraganglioma-pheochromocytoma (PGL/PCC) syndrome. This result does not rule out the presence of another alteration that may be responsible for the symptoms seen in this individual. Some individuals who have a diagnosis of PGL/PCC may have mutations or deletions that are not identified by the described testing methodology. In addition, up to 10% of individuals with a familial PGL/PCC phenotype may have mutations in genes other than SDHB, SDHC, and SDHD.					
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

* Report times for Mayo performed tests are CST/CDT

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SDH Deletion Detection		Performed			MCR

RECEIVED: **REPORTED:** 06/04/2013 18:13

* Performing Site:

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

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