

Patient Name SAMPLEREP,SDHDD	Patient ID SA00055256	Age 46	Gender F	Order # SA00055256
Ordering Phys UNKNOWN,PROVIDER				DOB 06/10/1966
Client Order # SA00055256	Account Information			Report Notes
Collected 03/15/2013 01:07	C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901			
Printed 06/21/2013 14:34				

Test	Flag	Results	Unit	Reference Value	Perform Site*
SDH Deletion Detection					
RECEIVED: 03/15/2013 01:07 REPORTED: 06/21/2013 14:00					
Reason for Referral		Not provided			MCR
Reason for referral was not provided by the requesting physician.					
Method		Deletion detection			MCR
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 large deletions were detected in SDHB, SDHC, or SDHD.					
Interpretation		Negative			MCR
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, a small percentage of individuals with a PGL/PCC phenotype may have mutations in genes other than SDHB, SDHC, and SDHD. If sequence analysis of the SDHB, SDHC, and SDHD genes has not been performed, it is recommended, since the majority of mutations that have been detected in these genes are identified via sequencing. Please contact the Endocrine Laboratory at 1-800-533-1710 or the online test catalog at mayomedicallaboratories.com for information about how to order sequence analysis for SDHB (89551/SDHSB), SDHC (89552/SDHSC), and/or SDHD (89553/SDHSD). For future reference, sequencing and deletion detection for the SDHB, SDHC, and SDHD genes are also available in a reflexive manner when SDH Full Gene Analysis is ordered (89550/SDHSP).					
Comment		GC consult			MCR
A genetic consultation may be of benefit.					
Reviewed by		LYNETTE HANSEN			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. Rarely, individuals may have a mutation or deletion in the gene(s)					

Performing Site Legend on Last Page of Report

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* Report times for Mayo performed tests are CST/CDT

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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:

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