

1-800-533-1710

PATIENT NAME SDH, TESTING		PATIENT NU L3MRNB99			<b>AGE</b> 48	SEX M	<b>ACCESSION #</b> B9962124
ORDERING PHYSICIAN		CLIENT ORD	DER#		•	•	ACCOUNT # LIAISONS
COLLECTION 08/22/11 01:11 P DATE TIME  Test Client Attn: Mayo Liaisons 200 First Street SW Rochester, MN 55905 507-284-8202	RECEIVED 08/22/11 ( DATE	REPORT PR 08/22/11 DATE	INTED 02:34 P TIME	SPECIMEN INFORM. DATE OF BIRTH:	ATION		

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LO **REF RANGE** PERFORM SITE \* **TEST REQUESTED** 

REPORTED: 08/22/11 01:31 P **DNA Extraction** 

Comment Genomic DNA was MCR

extracted.

SDH Subunit B Gene Analysis

REPORTED: 08/22/11 01:31 P Reason for Referral **MCR** 

Patient has clinical features suggestive of hereditary

paraganglioma-pheochromocytoma (PGL/PCC) syndrome. Test for the presence of mutation(s) within the SDHB gene.

Method **MCR** 

Fluorescent DNA sequence analysis was used to test for the presence of mutations in all eight exons and exon-intron boundaries of the SDHB gene.

Result MCR

This individual was shown to be heterozygous for the following mutation in the SDHB gene:

Exon 2, c.136C>T, p.Arg46X (p.R46X)

**MCR** Interpretation

This result is consistent with a diagnosis of PGL/PCC syndrome. Since a mutation has been identified in this individual, genetic testing for this specific mutation in other family members is recommended. Please contact the laboratory at 1-800-533-1710 or the online test catalog at mayomedicallaboratories.com for information about how to order SDH Known Mutation (89554) for sequencing mutations.

Comment **MCR** 

A genetic consultation may be of benefit.

Reviewed By **MCR** 

Stefan Grebe, MD, PhD

This test was developed and its performance characteristics determined by Laboratory Medicine and Pathology, Mayo Clinic. It has not been cleared or approved by the U.S. Food and Drug

\* Perform Site Legend on last page of report

PATIENT NAME	ORDER STATUS	COLLECTION DATE AND TIME
SDH, TESTING	Final	08/22/11 01:11 P



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DATE	TIME	DATE	TIME	DATE	TIME				
Test Clie	ent								
Attn: Ma	ayo Liaisons								
200 Firs	t Street SW								
Rochest	er, MN 55905								
507-284	-8202								

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TEST REQUESTED LO REF RANGE PERFORM SITE \*

Administration.

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 lab upon request.

Sequencing Performed MCR

## \* PERFORMING SITE

MCR Mayo Clinic Dpt of Lab Med & Pathology
Lab Director: Franklin R. Cockerill, III, M.D.
200 First Street SW Rochester, MN 55905

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SDH, TESTING	Final	08/22/11 01:11 P