MAYO CLINIC
Mayo Medical Laboratories

LABORATORY SERVICE REPORT

1-800-533-1710

PATIENT NAME	PATIENT NUMBER	AGE	SEX	ACCESSION #
SDH, TESTING	L3MRNB9962125	48	M	B9962125

ORDERING PHYSICIAN	CLIENT ORDER #	ACCOUNT #
		LIAISONS

COLLECTION DATE TIME	RECEIVED DATE TIME	REPORT PRINTED DATE TIME	SPECIMEN INFORMATION
08/22/11 01:12 P	08/22/11 01:12 P	08/22/11 02:34 P	DATE OF BIRTH:

Test Client
Attn: Mayo Liaisons
200 First Street SW
Rochester, MN 55905
507-284-8202

TEST REQUESTED	HI LO	REF RANGE	PERFORM SITE *

DNA Extraction — REPORTED: 08/22/11 01:39 P

Comment — Genomic DNA was extracted. — MCR

SDH Subunit C Gene Analysis — REPORTED: 08/22/11 01:39 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 SDHC gene.

Method — MCR

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

Result — MCR

This individual was shown to be heterozygous for the following mutation in the SDHC gene:
Exon 5, c.397C>T,p.R133X

Interpretation — MCR

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*

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SDH, TESTING	Final	08/22/11 01:12 P

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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 200 First Street SW Rochester, MN 55905	Lab Director: Franklin R. Cockerill, III, M.D.

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SDH, TESTING	Final	08/22/11 01:12 P