MAYO CLINIC
Mayo Medical Laboratories

LABORATORY SERVICE REPORT

1-800-533-1710

PATIENT NAME	PATIENT NUMBER	AGE	SEX	ACCESSION #
SDH, TESTING	L3MRNB9962121	48	M	B9962121

ORDERING PHYSICIAN	CLIENT ORDER #	ACCOUNT #
		C7999998

COLLECTION DATE TIME	RECEIVED DATE TIME	REPORT PRINTED DATE TIME	SPECIMEN INFORMATION
08/22/11 12:51 P	08/22/11 12:51 P	08/22/11 02:33 P	DATE OF BIRTH:

STUSTEST
Attn:
200 First Street SW
Rochester, MN 55901
507-266-5730

TEST REQUESTED	HI LO	REF RANGE	PERFORM SITE *
DNA Extraction		**REPORTED: 08/22/11 01:02 P**	
Comment	**Genomic DNA was extracted.**		**MCR**
SDH Gene Analysis		**REPORTED: 08/22/11 01:03 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, SDHC, and SDHD genes.			
Method			**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			**MCR**
This individual was shown to be heterozygous for the following mutation in the SDHD gene: Exon 3, c.242C>T, p.Pro81Leu (p.P81L)			
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 further information about how to order SDH Known Mutation (89554) for sequencing mutations or SDH Deletion Detection (89555) for large deletions of one or more exons.			
Comment			**MCR**
A genetic consultation may be of benefit.			
Reviewed By			**MCR**
Stefan Grebe, MD, PhD			

** Perform Site Legend on last page of report*

PATIENT NAME	ORDER STATUS	COLLECTION DATE AND TIME
SDH, TESTING	Final	08/22/11 12:51 P

Specimen receipt and report times are in CST/CDT

REPRINT

PATIENT NAME	PATIENT NUMBER	AGE	SEX	ACCESSION #
SDH, TESTING	L3MRNB9962121	48	M	B9962121

ORDERING PHYSICIAN	CLIENT ORDER #	ACCOUNT #
		C7999998

COLLECTION DATE TIME	RECEIVED DATE TIME	REPORT PRINTED DATE TIME	SPECIMEN INFORMATION
08/22/11 12:51 P		08/22/11 02:33 P	DATE OF BIRTH:

STUSTEST
Attn:
200 First Street SW
Rochester, MN 55901
507-266-5730

TEST REQUESTED	HI LO	REF RANGE	PERFORM SITE *

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.