

1-800-533-1710

PATIENT NAME TESTING, QA89015		PATIENT NUMBER		AGE 33	SEX F	ACCESSION # G9134773
ORDERING PHYSICIAN		CLIENT ORDER #			ACCOUNT # LIAISONS	
COLLECTION 09/15/09 02:23 P DATE TIME	RECEIVED 09/15/09 02:23 P DATE TIME	REPORT PRINTED 11/11/09 01:29 P DATE TIME		SPECIMEN INFORMATION 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 *
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NPC Mutation Screen

Specimen	Blood	MCR
Specimen ID	1033332	MCR
Order Date	05 Oct 2009 11:36	MCR
Reason For Referral	Not Provided.	MCR
Method		MCR

DNA sequence analysis was performed to test for the presence of a mutation in all 25 exons of the NPC1 gene and all 5 exons of the NPC 2 gene. Gene dosage analysis (MLPA) was used to test for the presence of large deletions, duplications, and other large genomic rearrangements within these genes.

Result **MCR**

A mutation was NOT detected.

Interpretation **MCR**

In the absence of biochemical evidence of disease, these results decrease the likelihood, but do not rule out the diagnosis of Niemann-Pick type C (NPC) for this individual. Other types of mutations that are not detectable by the methods described above, such as promoter or intronic mutations, may be present in the NPC1 or NPC2 genes. Based on a detection rate of 85% for NPC1 and 90-95% for NPC2, approximately 1-2% of patients with NPC are predicted to have an unidentified mutation on both chromosomes. Because phenotypic overlap is occasionally observed among the different types of lysosomal storage diseases, laboratory testing including cholesterol esterification and filipin staining on a fibroblast sample of the affected individual is recommended to establish a diagnosis of NPC. A list of common polymorphisms identified for this patient is available upon request.

* Perform Site Legend on last page of report

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A genetic consultation may be of benefit.

CAUTIONS:

Test results should be interpreted in context of clinical findings, family history, and other laboratory data.

Misinterpretation of results may occur if the information provided is inaccurate or incomplete.

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. Bone marrow transplants from allogenic donors will interfere with testing. Call Mayo Medical Laboratories for instructions for testing patients who have received a bone marrow transplant.

MLPA Performed?	Yes.	MCR
See #89851, NPC Large Deletion/Duplication, MLPA, for billing information.		
Reviewed BY	Keri Jane Kruckeberg	MCR
Release Date	05 Oct 2009 11:38	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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