Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,NPCMS N	SA00058852	46	F	SA00058852
Ordering Phys CLIENT,CLIENT				**DOB** 06/10/1966
Client Order # SA00058852	**Account Information**			**Report Notes**
Collected 06/06/2013 00:00	C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901			
Printed 06/11/2013 16:04				

Test	Flag	Results	Unit	Reference Value	Perform Site*
NPC Mutation Screen					
RECEIVED: 06/07/2013 15:47 **REPORTED:** 06/10/2013 16:28					
Specimen		Blood			MCR
Specimen ID		1062119			MCR
Order Date		10 Jun 2013 09:14			MCR
Reason For Referral					MCR

Possible diagnosis of Niemann-Pick disease type C (NPC). Test for the presence of mutations in the NPC1 and NPC2 genes.

Method MCR

Bi-directional sequence analysis was performed to test for the presence of mutations in all coding regions and exon/intron boundaries of the NPC1 gene (GenBank accession number; NM_000271.4) and NPC2 gene (GenBank accession number; NM_006432.3). Gene dosage analysis (MLPA) was used to test for the presence of large deletions and duplications in these genes.

Result MCR

A mutation was NOT detected in NPC1 or NPC2.

Interpretation MCR

This result decreases the likelihood but does not exclude the diagnosis of NPC. We predict that there may be disease causing mutations in the NPC1 or NPC2 genes not identifiable by the methods described (e.g. mutations in the promoter or deep intronic mutations). This assay does not rule out the presence of mutations in other genes that are associated with metabolic disease.

This result should be interpreted in the context of clinical findings, family history, and other laboratory data (e.g. cholesterol esterification and filipin staining on a fibroblast sample).

A genetic consultation may be of benefit.

Unless reported or predicted to cause disease, alterations found deep in the intron or alterations that do not result in an amino acid substitution are not reported. These and common polymorphisms identified for this patient are available upon request.

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

Performing Site Legend on Last Page of Report

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Test	Flag	Results	Unit	Reference Value	Perform Site*
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. Laboratory developed test.					
MLPA Performed?		Yes.			MCR
Reviewed By Emily Christine Lauer					MCR
Release Date		10 Jun 2013 16:26			MCR

* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director: Franklin R. Cockerill, III, M.D.

* Report times for Mayo performed tests are CST/CDT

TEST SITE