Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,NPCKM A	SA00058851	46	F	SA00058851
Ordering Phys CLIENT,CLIENT				**DOB** 06/10/1966
Client Order # SA00058851	**Account Information**			**Report Notes**
Collected 06/06/2013 00:00	C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901			
Printed 06/11/2013 16:03				

Test	Flag	Results	Unit	Reference Value	Perform Site*
NPC Known Mutation					
RECEIVED: 06/07/2013 15:42 **REPORTED:** 06/10/2013 16:26					
Specimen		Blood			MCR
Specimen ID		1062118			MCR
Order Date		10 Jun 2013 09:13			MCR
Reason For Referral					MCR

Family history of Niemann-Pick disease type C (NPC). Test for the presence of familial alterations in the NPC1 gene.

Method MCR

DNA sequence analysis was used to test for the presence of the p.I1061T (c.3182T>C) alteration in exon 21 in the NPC1 gene (GenBank accession number; NM_000271.4). Analysis for this specific alteration was performed because it was identified in a family member.

Result MCR

The following heterozygous sequence change in NPC1 was detected:
Exon: 21
DNA change: c.3182T>C
Amino Acid change: p.I1061T (Ile1061Thr)
Classification: DELETERIOUS

Interpretation MCR

This alteration is a known deleterious mutation.

This result indicates that this individual is a carrier of NPC. This interpretation assumes that this individual is healthy and not clinically affected with NPC.

Since a mutation has been identified in the NPC1 gene, genetic testing of at risk family members is possible. If appropriate, genetic testing should be offered to this individual's reproductive partner to clarify their risk of having a child with NPC.

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

Performing Site Legend on Last Page of Report

TEST SITE

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,NPCKM A	SA00058851	46	F	SA00058851
Ordering Phys CLIENT,CLIENT				**DOB** 06/10/1966
Client Order # SA00058851	**Account Information**			**Report Notes**
Collected 06/06/2013 00:00	C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901			
Printed 06/11/2013 16:03				

Test	Flag	Results	Unit	Reference Value	Perform Site*
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.					
Laboratory developed test.					
Reviewed By					MCR
Emily Christine Lauer					
Release Date		10 Jun 2013 16:24			MCR

* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director:

TEST SITE