Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,NPCKM N	SA00058850	46	F	SA00058850
Ordering Phys CLIENT,CLIENT				**DOB** 06/10/1966
Client Order # SA00058850	**Account Information**			**Report Notes**
Collected 06/06/2013 00:00	C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901			
Printed 06/11/2013 16:02				

Test	Flag	Results	Unit	Reference Value	Perform Site*
NPC Known Mutation					
RECEIVED: 06/07/2013 15:41 **REPORTED:** 06/10/2013 16:24					
Specimen		Blood			MCR
Specimen ID		1062117			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 p.I1061T alteration in the NPC1 gene was NOT detected.					
Interpretation					MCR
Absence of the mutation(s) previously identified in a family member indicates that this individual is at no greater risk than someone in the general population to be a carrier of NPC.					
This assay does not rule out the presence of other disease causing mutations in the NPC1 or NPC2 genes, or other genes associated with metabolic disease. Errors in the diagnosis or pedigree provided to us, including non paternity, may lead to an erroneous interpretation of the test results.					
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. 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					

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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 Emily Christine Lauer					MCR
Release Date		10 Jun 2013 16:22			MCR

* Performing Site:

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