Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,MP3BK	SA00057507	37	F	SA00057507
Ordering Phys CLIENT,CLIENT				**DOB** 06/18/1975
Client Order # SA00057507	**Account Information**			**Report Notes**
Collected 05/17/2013 08:26	C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901			
Printed 06/11/2013 15:16				

Test	Flag	Results	Unit	Reference Value	Perform Site*

MPS IIIB, Known Mutation
RECEIVED: **REPORTED:** 05/23/2013 10:53

Reason For Referral — MCR

Family history of mucopolysaccharidosis type IIIB (Sanfilippo syndrome type B). Test for the presence of familial alterations in the NAGLU gene.

Result — MCR

The following heterozygous intronic sequence change was detected:
Intron: 4
DNA change: c.764+1G>A
Classification: LIKELY DELETERIOUS

Interpretation — MCR

This alteration alters a canonical splice site and is therefore predicted to be deleterious.

The presence of a mutation previously identified in a family member indicates that this individual is a carrier of mucopolysaccharidosis type IIIB (MPS-IIIB), also known as Sanfilippo syndrome type B. This interpretation assumes that this individual is healthy and is not clinically affected with MPS-IIIB.

Since a mutation has been identified in the NAGLU gene, genetic testing of at risk family members is possible. If appropriate, enzymatic and molecular testing should be offered to this individual's reproductive partner to further clarify their risk of having a child with MPS-IIIB.

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

Performing Site Legend on Last Page of Report

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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.					
Method					MCR
DNA sequence analysis was used to test for the presence of the c.764+1G>A and c.1022-1G>C alterations in introns 4 and 5, respectively, in the NAGLU gene. Testing was performed for these specific alterations because they were previously identified in a family member of this individual. Mutation nomenclature is based on GenBank accession number NM_000263.3.					
Specimen		Blood			MCR
Reviewed By					MCR
Dimitar Gavrilov MD, PhD					
Release Date		23 May 2013 10:52			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.