Patient Name	Patient ID	Age	Gender	Order #
TESTINGRNV,REPORTS	SA00063722	21D	F	SA00063722
Ordering Phys CLIENT,CLIENT				**DOB** 10/01/2013
Client Order # SA00063722	**Account Information**			**Report Notes**
Collected 10/22/2013 00:00	C7028846-DLMP Rochester SDSC 2 - Client Support Rochester, MN 55901			
Printed 11/13/2013 15:33				

Test	Flag	Results	Unit	Reference Value	Perform Site*

MPS IIIA, Full Gene Analysis
RECEIVED: 10/22/2013 14:07 **REPORTED:** 10/30/2013 10:26

Reason for Referral MCR

Patient reported to have features suggestive of mucopolysaccharidosis type IIIA (Sanfilippo syndrome type A; sulfamidase deficiency). Test for the presence of mutations in the SGSH gene.

Result MCR

A mutation was NOT detected.

Interpretation MCR

This result decreases the likelihood but does not completely exclude a diagnosis of mucopolysaccharidosis type IIIA (MPS-IIIA), also known as Sanfilippo syndrome type A or sulfamidase deficiency.

We predict that there may be disease causing mutations in the SGSH gene not identifiable by the method described (e.g., large deletions/duplications, promoter mutations, or deep intronic mutations).

This assay does not rule out the presence of disease causing mutations in other genes associated with metabolic disease. This result should be interpreted in the context of clinical findings, family history, and other laboratory testing (e.g. urinary mucopolysaccharides and sulfamidase activity).

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 the context of clinical findings, family history, and other laboratory data. Errors in our interpretation of results may occur if information given is inaccurate or incomplete.

Rare polymorphisms exist that could lead to false-negative or false-positive results. If results obtained do not match the clinical findings, additional testing should be considered.

Performing Site Legend on Last Page of Report

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TESTINGRNV,REPORTS	SA00063722	21D	F	SA00063722
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Test	Flag	Results	Unit	Reference Value	Perform Site*
A previous bone marrow transplant from an allogenic donor 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
Bi-directional sequence analysis was performed to test for the presence of mutations in all coding regions and intron/exon boundaries of the SGSH gene. Mutation nomenclature is based on GenBank accession number NM_000199.3.					
Specimen		Blood			MCR
Reviewed By		Devin Oglesbee PhD			MCR
Release Date		30 Oct 2013 10:23			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.