MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
TESTINGRNV,REPORTS	SA00063723	21D	F	SA00063723
Ordering Phys CLIENT,CLIENT				**DOB** 10/01/2013
Client Order # SA00063723	**Account Information**			**Report Notes**
Collected 10/22/2013 00:00	C7028846-DLMP Rochester SDSC 2 - Client Support Rochester, MN 55901			
Printed 11/13/2013 15:34				

Test	Flag	Results	Unit	Reference Value	Perform Site*

MPS IIIA, Full Gene Analysis
RECEIVED: 10/22/2013 14:08 **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

The following homozygous deletion was detected:
Exon: 8
DNA change: c.1080delC
Amino Acid change: p.V361SfsX52 (Val361SerfsX52)
Classification: DELETERIOUS

Interpretation MCR

The c.1080delC alteration is a known deleterious mutation.

This result is consistent with a diagnosis of mucopolysaccharidosis type IIIA (MPS-IIIA), also known as Sanfilippo syndrome type A or sulfamidase deficiency.

Correlation of these results with biochemical and clinical findings is recommended.

Since mutations have been identified, testing of at risk family members is possible. Mutation-specific testing for MPS-IIIA is available at Mayo Medical Laboratories by ordering MP3AK/61821 MPS IIIA, Known Mutation. Please contact the Molecular Genetics Laboratory at 1-800-533-1710 with questions about this test.

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.

Performing Site Legend on Last Page of Report

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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.					
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: