

Laboratory Service Report

1-800-533-1710

MCR

MCR

MCR

Patient Name	Patient ID	Age		Order #
TESTINGRNV,REPORTS	SA00063725	21D	F	SA00063725
Ordering Phys				DOB
CLIENT,CLIENT				10/01/2013
Client Order # SA00063725	Account Information			Report Notes
Collected	C7028846-DLMP Rock			
10/22/2013 00:00	SDSC 2 - Client Suppo	ort		
Printed	Rochester, MN 55901			
11/13/2013 15:39				

Test Flag Results Unit Value Site*

MPS IIIA, Known Mutation

RECEIVED: 10/22/2013 14:10 **REPORTED:** 10/30/2013 10:28

Reason for Referral
Family history of mucopolysaccharidosis type IIIA

(Sanfilippo syndrome type A; sulfamidase deficiency). Test

for the presence of familial alterations in the SGSH gene. Result $\,$

The following homozygous deletion was detected:

Exon: 8

DNA change: c.1080delC

Amino Acid change: p.V361SfsX52 (Val361SerfsX52)

Classification: DELETERIOUS

Interpretation

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

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

Since mutations have been identified in the SGSH gene, genetic testing of at risk family members is possible.

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 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.

Performing Site Legend on Last Page of Report

Patient Name	Collection Date and Time	Report Status
TESTINGRNV,REPORTS	10/22/2013 00:00	Final
Page 1 of 2		>> Continued on Next Page >>

^{*} Report times for Mayo performed tests are CST/CDT



Laboratory Service Report

1-800-533-1710

Patient Name TESTINGRNV,REPORTS	Patient ID SA00063725	Age 21D	Gender F	Order # SA00063725
Ordering Phys CLIENT,CLIENT		•	•	DOB 10/01/2013
Client Order # SA00063725	Account Information			Report Notes
Collected 10/22/2013 00:00	SDSC 2 - Client Suppo	C7028846-DLMP Rochester SDSC 2 - Client Support		
Printed 11/13/2013 15:39	Rochester, MN 55901			

Test	Flag Results	Unit	Reference Value	Perform Site*
of the p.V361SfsX52 (c. SGSH gene. Testing was alteration because it w	as performed to test for the pres 1080delC) alteration in exon 8 in performed for this specific as previously in a family member on nomenclature is based on GenBa	of		MCR
Specimen Reviewed By Release Date	Blood Devin Oglesbee P 30 Oct 2013 10:2			MCR MCR 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.

Patient Name	Collection Date and Time	Report Status
TESTINGRNV,REPORTS	10/22/2013 00:00	Final
Page 2 of 2		** End of Report **

^{*} Report times for Mayo performed tests are CST/CDT