

Laboratory Service Report

1-800-533-1710

REPORTED 02/20/2013 17:14

Patient Name TESTING,REPORTS	Patient ID SA00054083	Age 16D	Gender M	Order # SA00054083	
Ordering Phys CLIENT, CLIENT	SA00034063	100	IVI	DOB 02/04/2013	
Client Order # SA00054083	Account Information			Report Notes	
Collected 02/20/2013	3050 Superior Drive	· • • • • • • • • • • • • • • • • • • •			
Printed 03/06/2013 15:45	Rochester, MN 55901	Rochester, MN 55901			

Reference Perform
Test Flag Results Unit Value Site*

Hurler Syndrome, Known Mutation

Reason for Referral

MCR

Family history of mucopolysaccharidosis type I (Hurler/Scheie). Test for the presence of familial alterations in the IDUA gene.

Result

MCR

The p.W402X alteration was NOT detected.

Interpretation

MCR

Absence of the mutation(s) previously identified in an affected family member indicates that this individual is at no greater risk than someone in the general population to be a carrier of mucopolysaccharidosis type I (MPS-I). MPS-I includes into Hurler syndrome, Scheie syndrome, and Hurler-Scheie syndrome.

This assay does not rule out the presence of other disease causing mutations in this gene 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 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
TESTING,REPORTS	02/20/2013	Final
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^{*} Report times for Mayo performed tests are CST/CDT



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TESTING,REPORTS	SA00054083	16D	M	SA00054083	
Ordering Phys CLIENT,CLIENT			•	DOB 02/04/2013	
Client Order # SA00054083	Account Information			Report Notes	
Collected 02/20/2013	3050 Superior Drive	C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901			
Printed 03/06/2013 15:45	Rochester, MN 55901				

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Test	Flag	Results	Unit	Reference Value	Perform Site*
Laboratory developed test					
Method					MCR
DNA sequence analysis was	performed to t	test for the	presence		
of the p.W402X (c.1205G>A)) alteration in	n exon 9 in t	he IDUA		
gene. Testing was perform	-				
because it was previously		-			
this individual. Mutation		is based on	GenBank		
accession number NM 000203					
Specimen		Blood			MCR
Reviewed By					MCR
Dimitar Gavrilov MD, PhD					
Release Date		20 Feb 2013	17:13		MCR

* Performing Site:

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

Patient Name	Collection Date and Time	Report Status
TESTING,REPORTS	02/20/2013	Final
Page 2 of 2		** End of Report **

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