

Patient Name SAMPLEREP,MCIV	Patient ID SA00007371	Age 32	Gender F	Order # SA00007371
Ordering Phys				DOB 04/04/1980
Client Order # SA00007371	Account Information			Report Notes
Collected 07/31/2012 13:00	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 08/15/2012 14:12				

Test	Flag	Results	Unit	Reference Value	Perform Site*
Mucopolipidosis IV, Mutation Analysis				REPORTED 08/14/2012 16:59	
Specimen		Blood			MCR
Specimen ID		976470			MCR
Order Date		01 Aug 2012 13:08			MCR
Reason For Referral		Carrier screen for mucopolipidosis IV. Test for the presence of a mutation in the MCOLN1 gene.			MCR
Method		A PCR-based assay was used to test for the following mutations in the MCOLN1 gene: IVS3(-2)A>G and delta6.4kb.			MCR
Result		One copy of the delta6.4kb mutation was identified.			MCR
Interpretation		This result indicates that this individual is a carrier of mucopolipidosis IV. This interpretation assumes that this individual is healthy and is not clinically affected with mucopolipidosis IV.			MCR
		Since a mutation has been identified, genetic testing of at risk family members is possible. If appropriate, genetic testing should be offered to this individual's reproductive partner.			
		A genetic consultation may be of benefit.			
		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.			
		Laboratory developed test.			
Reviewed By:		D Brian Dawson PhD			MCR
Release Date		14 Aug 2012 16:58			MCR

Performing Site Legend on Last Page of Report

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* Report times for Mayo performed tests are CST/CDT

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>> Accession SA00007371 - Continued From Previous Page <<
>> Do Not Discard <<

* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director:
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Patient Name SAMPLEREP,MCIV	Collection Date and Time 07/31/2012 13:00	Report Status Final
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