

Mila

Laboratory Service Report

1-800-533-1710

Patient Name SAMPLEREPORT,MCIV	Patient ID SA00007370	Age 32	Gender F	Order # SA00007370
Ordering Phys				DOB 04/04/1980
Client Order # SA00007370	Account Information			Report Notes
Collected 07/31/2012	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE			
Printed 08/15/2012 14:11	ROCHESTER,MN 559	901		

Test	Flag Resu	ts Uni	it	Reference Value	Perform Site*
ucolipidosis IV, Mutation Analysi	s	RE	EPORTED 08/14/2	2012 16:59	
Specimen	Blood	l			MCR
Specimen ID	97646	59			MCR
Order Date	01 At	ıg 2012 13:08			MCR
Reason For Referral					MCR
Carrier screen for mucolip	idosis IV. Test f	or the presence			
of a mutation in the MCOLN	1 gene.				
Method					MCR
A PCR-based assay was used	to test for the f	ollowing			
mutations in the MCOLN1 ge	ne: IVS3(-2)A>G and	d delta6.4kb.			
Result					MCR
Neither of the listed muta	tions was detected	•			
Interpretation					MCR
Having excluded the presen					
risk for this individual t					
mucolipidosis IV mutation					
risk assumes that there is					
mucolipidosis IV. Additio	nally, this calcul	ation is based			

Because there is little information about the carrier frequency and mutation detection rate for individuals of non-Ashkenazi Jewish ancestry, we are unable to provide a specific revised risk assessment for individuals of other ethnicities.

on a mutation detection rate of 95% (for Ashkenazi Jewish ancestry) and a population carrier frequency of 1/127.

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.

Performing Site Legend on Last Page of Report

regression of Edge of Report				
Patient Name	Collection Date and Time	Report Status		
SAMPLEREPORT,MCIV	07/31/2012	Final		
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Test Flag Results Unit Reference Value Site*

Laboratory developed test.

Reviewed By:
Release Date

D Brian Dawson PhD
Release Date

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
SAMPLEREPORT.MCIV	07/31/2012	Final
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