

Patient Name TESTING,REPORTS	Patient ID SA00065080	Age 8D	Gender F	Order # SA00065080
Ordering Phys CLIENT,CLIENT				DOB 01/02/2014
Client Order # SA00065080	Account Information			Report Notes
Collected 01/10/2014 00:00	C7028846-DLMP Rochester SDSC 2 - Client Support Rochester, MN 55901			
Printed 02/05/2014 14:59				

Test	Flag	Results	Unit	Reference Value	Perform Site*
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GNPTAB Gene, Known Mutation
RECEIVED: 01/10/2014 14:50 **REPORTED:** 01/17/2014 18:33

Reason for Referral

MCR

Family history of GNPTAB-related Mucolipidoses. Test for the presence of mutations in the GNPTAB gene.

Result

MCR

The p.Q845STOP mutation 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 GNPTAB-related Mucolipidoses.

This assay does not rule out the presence of other disease-causing mutations in this gene or other genes associated with GNPTAB-related Mucolipidoses, or other lysosomal diseases. Errors in the diagnosis or pedigree provided to us, including non-paternity, may lead to an erroneous interpretation of 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 TESTING,REPORTS	Collection Date and Time 01/10/2014 00:00	Report Status Final
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* Report times for Mayo performed tests are CST/CDT

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For research use only.					
Method		DNA sequence analysis was performed to test for the presence of the p.Q845STOP (c.2533C>T) alteration in exon 13 of the GNPTAB gene (GenBank accession number NM_024312.4).			MCR
Specimen		Blood			MCR
Reviewed By		Devin Oglesbee PhD			MCR
Release Date		17 Jan 2014 18:29			MCR

* Performing Site:

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