MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,BLM	SA00007358	32	F	SA00007358
Ordering Phys				**DOB** 04/04/1980
Client Order # SA00007358	**Account Information**			**Report Notes**
Collected 07/31/2012 13:00	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 08/15/2012 14:03				

Test	Flag	Results	Unit	Reference Value	Perform Site*
Bloom Syndrome, Mutation Analysis			REPORTED 08/14/2012 16:52		
Specimen		Blood			MCR
Specimen ID		976455			MCR
Order Date		01 Aug 2012 13:04			MCR
Reason For Referral					MCR

Carrier screen for Bloom syndrome. Test for the presence of a mutation in the BLM gene.

Method MCR

A PCR-based assay was used to test for the 2281del6/ins7 mutation in the BLM gene.

Result MCR

The 2281del6/ins7 mutation was NOT detected.

Interpretation MCR

Having excluded the presence of the 2281del6/ins7 mutation, the risk for this individual to be a carrier of another Bloom syndrome mutation is approximately 1/10,601. This risk assumes that there is no family history of Bloom syndrome. Additionally, this calculation is based on a mutation detection rate of 99% (for Ashkenazi Jewish ancestry) and a population carrier frequency of 1/107.

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.

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

* Report times for Mayo performed tests are CST/CDT

TEST SITE

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Test	Flag	Results	Unit	Reference Value	Perform Site*
Laboratory developed test.					
Review By:		D Brian Dawson PhD			MCR
Release Date		14 Aug 2012 16:47			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
SAMPLEREPORT,BLM	07/31/2012 13:00	Final
Page 2 of 2		** End of Report **

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TEST SITE