

Patient Name SAMPLEREP, BLM	Patient ID SA00007359	Age 32	Gender F	Order # SA00007359
Ordering Phys				DOB 04/04/1980
Client Order # SA00007359	Account Information			Report Notes
Collected 07/31/2012 13:00	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER, MN 55901			
Printed 08/15/2012 14:04				

Test	Flag	Results	Unit	Reference Value	Perform Site*
Bloom Syndrome, Mutation Analysis			REPORTED	08/14/2012 16:52	
Specimen		Blood			MCR
Specimen ID		976456			MCR
Order Date		01 Aug 2012 13:04			MCR
Reason For Referral		Carrier screen for Bloom syndrome. Test for the presence of a mutation in the BLM gene.			MCR
Method		A PCR-based assay was used to test for the 2281del6/ins7 mutation in the BLM gene.			MCR
Result		One copy of the 2281del6/ins7 mutation was identified.			MCR
Interpretation		This result indicates that this individual is a carrier of Bloom syndrome. This interpretation assumes that this individual is healthy and is not clinically affected with Bloom syndrome.			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.			
Review By:		D Brian Dawson PhD			MCR
Release Date		14 Aug 2012 16:47			MCR

Performing Site Legend on Last Page of Report

Patient Name SAMPLEREP, BLM	Collection Date and Time 07/31/2012 13:00	Report Status Final
Page 1 of 2		>> Continued on Next Page >>

* Report times for Mayo performed tests are CST/CDT

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>> Accession SA00007359 - 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 SAMPLEREPORT,BLM	Collection Date and Time 07/31/2012 13:00	Report Status Final
Page 2 of 2		** End of Report **

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