

Patient Name SAMPLEREP,FBNN A	Patient ID SA00062364	Age 47	Gender F	Order # SA00062364
Ordering Phys CLIENT,CLIENT				DOB 06/10/1966
Client Order # SA00062364	Account Information		Report Notes	
Collected 09/15/2013 00:00	C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901			
Printed 09/17/2013 07:26				

Test	Flag	Results	Unit	Reference Value	Perform Site*
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Neonatal Marfan Syndrome, FBN1 Gene
RECEIVED: 09/16/2013 08:43 **REPORTED:** 09/16/2013 18:00

Neonatal Marfan Syndrome, FBN1 Gene

FBN1 Neonatal Result

MCR

One copy of the following mutation was detected in FBN1:
 Exon 26, nucleotide c.3257G>A, amino acid p.Cys1086Tyr
 (p.C1086Y).

FBN1 Neonatal Interpretation

MCR

This FBN1 mutation is consistent with the neonatal Marfan syndrome features observed in this patient. Appropriate surveillance and management strategies should be considered.

Mutations associated with neonatal Marfan syndrome are usually de novo in the affected individual, but can sometimes be due to parental germline mosaicism. If parental germline mosaicism is present, it may or may not be detectable due to limitations in current testing technology. If desired, please contact the laboratory at 1-800-533-1710 or the on-line test catalog at www.mayomedicallaboratories.com for information about how to order the test for FBN1 Gene, Known Mutation (89311). Fluorescent DNA sequence analysis was used to test for the presence of mutations in exons 24-33 and flanking regions of the FBN1 gene (GenBank accession number NM_00138.3).

A genetic consultation may be of benefit.

A list of common polymorphisms identified for this patient is available from the lab upon request.

CAUTIONS:

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.

Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.

Bone marrow transplants from allogenic donors will interfere with testing. Call Mayo Medical Laboratories for instructions for testing patients who have received a bone

Performing Site Legend on Last Page of Report

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Page 1 of 2		>> Continued on Next Page >>

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marrow transplant. Laboratory developed test. Reviewed By FBN1 Neonatal Screen Sequencing		Jamie Bruflat Performed			MCR MCR

* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director:
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Page 2 of 2		** End of Report **

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