MAYO CLINIC
Mayo Medical Laboratories

1-800-533-1710

FBNN

FBN1, Partial Gene Sequence, Neonatal Marfan Syndrome

Patient ID	Patient Name		Birth Date	Gender	Age
SA00062363	SAMPLEREPORT, FBNN N		1966-06-10	F	47
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00062363	SA00062363	Client, Client			
Account Information		**Collected**			
C7028846 DLMP Rochester		15 Sep 2013 00:00			

Neonatal Marfan Syndrome, FBN1 Gene

Neonatal Marfan Syndrome, FBN1 Gene

FBN1 Neonatal Result MCR

A mutation was not detected in FBN1.

FBN1 Neonatal Interpretation MCR

This result does not rule out the diagnosis of Marfan Syndrome. Individuals presenting in the neonatal period with severe Marfan syndrome (also known as Neonatal Marfan syndrome) have been shown to harbor mutations in exons 24 to 32 of the FBN1 gene. However, some individuals who have a diagnosis of Marfan Syndrome and involvement of the FBN1 gene may have mutations that are not identified by the described testing methodology. Testing of the remainder of the exons of the FBN1 gene is recommended if there is a high suspicion for the diagnosis of Marfan syndrome. Please contact the laboratory at 1–800–533–1710 or the on-line test catalog at mayomedicallaboratories.com for information about how to order the test for FBN1, Full Gene Sequence (89308).

ADDITIONAL INFORMATION

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.

If the patient has had an allogeneic blood or marrow transplant or a recent (i.e. less than 6 weeks from time of sample collection) heterologous blood transfusion these results may be inaccurate due to the presence of donor DNA. Laboratory developed test.

Reviewed By MCR

Jamie Bruflat

FBN1 Neonatal Screen Sequencing MCR

Performed

Received: 16 Sep 2013 08:43 **Reported:** 16 Sep 2013 17:59

Performing Site Legend

Code	Laboratory	Address
MCR	Mayo Clinic Dept. of Lab Med and Pathology	200 First Street SW, Rochester, MN 55905