

Patient ID SA00059587	Patient Name SAMPLEREPORT, FBN1 N	Birth Date 1966-06-10	Gender F	Age 47
Order Number SA00059587	Client Order Number SA00059587	Ordering Physician Client, Client	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 30 Jun 2013 00:00		

FBN1, Full Gene Sequence

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FBN1 Full Gene Result

MCR

A mutation was not detected in FBN1.

A genetic consultation may be of benefit.

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

FBN1 Full Gene Interpretation

MCR

This result does not rule out the diagnosis of Marfan syndrome or other FBN1-associated syndrome. Some individuals with features of Marfan syndrome or other FBN1-associated syndrome and involvement of the FBN1 gene may have a mutation that is not identified by the described testing methodology. In addition, some individuals with this phenotype may have a mutation in a gene other than FBN1, including TGFBR2 and TGFBR1. If applicable, 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 tests for TGFBR2 Full Gene Sequence (89461) and/or TGFBR1, Full Gene Sequence (89459).

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.

ADDITIONAL INFORMATION

Fluorescent DNA sequence analysis was used to test for the presence of mutations in all 65 exons and exon-intron boundaries of the FBN1 gene (GenBank accession number NM_00138.3).

Reviewed By

MCR

Jamie Bruflat

FBN1, Full Gene Sequencing

MCR

Performed

Received: 03 Jul 2013 13:37

Reported: 16 Sep 2013 17:56

Performing Site Legend

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