

Patient ID <b>SA00059695</b>	Patient Name <b>SAMPLEREPOR, FBN1 N</b>	Birth Date <b>1966-06-10</b>	Gender <b>F</b>	Age <b>47</b>
Order Number <b>SA00059695</b>	Client Order Number <b>SA00059695</b>	Ordering Physician <b>Client, Client</b>	Report Notes	
Account Information <b>C7028846 DLMP Rochester</b>		Collected <b>02 Jul 2013 00:00</b>		

## FBN1, Full Gene Sequence

### FBN1, Full Gene Sequence

#### FBN1 Full Gene Result

MCR

One copy of the following mutation was detected in FBN1:

Exon 29, nucleotide c.3670C>T, amino acid p.Gln1224X (p.Q1224X).

#### FBN1 Full Gene Interpretation

MCR

This result is consistent with a disease-causing FBN1 mutation. Appropriate surveillance and management strategies should be considered.

Since a mutation has been identified in the FBN1 gene in this individual, genetic testing for this specific mutation in other family members is recommended. Please contact the laboratory at 1-800-533-1710 or the on-line test catalog at [www.mayomedicallaboratories.com](http://www.mayomedicallaboratories.com) for information about how to order the test for FBN1 Gene, Known Mutation (89311).

#### 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).

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, Full Gene Sequencing

MCR

Performed

**Received:** 03 Jul 2013 13:37

**Reported:** 16 Sep 2013 17:58

#### Performing Site Legend

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