MAYO CLINIC
Mayo Medical Laboratories

1-800-533-1710

FBKM

FBN1 Genetic Analysis, Known Mutation

Patient ID	Patient Name		Birth Date	Gender	Age
SA00059586	SAMPLEREPORT, FBKM A		1966-06-10	F	47
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00059586	SA00059586	Client, Client			
Account Information		**Collected**			
C7028846 DLMP Rochester		30 Jun 2013 00:00			

FBN1 Gene, Known Mutation

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FBN1 Known Mut Result

MCR

One copy of the FBN1 Exon 52, nucleotide c.6495_6496insA, amino acid p.Asp2166ArgfsX3 familial mutation was detected in this individual.

FBN1 Known Mut Interpretation

MCR

The FBN1 c.6495_6496insA, p.Asp2166ArgfsX3 mutation was previously identified in a family member with features of Marfan syndrome (MFS). The presence of this mutation, therefore, suggests that this individual is at risk for development and/or exacerbation of features of MFS. Appropriate surveillance procedures and/or 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 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 a specific mutation in the FBN1 gene (GenBank accession number NM_00138.3), which was previously identified in an affected family member of this individual.

A genetic consultation may be of benefit.

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 Known Mutation Sequencing

MCR

Performed

Received: 03 Jul 2013 13:36 **Reported:** 16 Sep 2013 17:55

Performing Site Legend

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