

Patient ID <b>SA00059585</b>	Patient Name <b>SAMPLEREPORT, FBKM N</b>	Birth Date <b>1966-06-10</b>	Gender <b>F</b>	Age <b>47</b>
Order Number <b>SA00059585</b>	Client Order Number <b>SA00059585</b>	Ordering Physician <b>Client, Client</b>	Report Notes	
Account Information <b>C7028846 DLMP Rochester</b>		Collected <b>30 Jun 2013 00:00</b>		

## FBN1 Gene, Known Mutation

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#### FBN1 Known Mut Result

MCR

The FBN1 Exon 15, nucleotide c.1879C>T, amino acid p.Arg627Cys (p.R627C) familial mutation was not detected in this individual.

#### FBN1 Known Mut Interpretation

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The FBN1 p.R627C mutation was previously identified in a family member with features of Marfan syndrome. Since this mutation was not detected in this individual, this suggests that this patient is at no greater risk than someone in the general population for having Marfan syndrome.

#### 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 Brufat

#### FBN1 Known Mutation Sequencing

MCR

Performed

**Received:** 03 Jul 2013 13:35

**Reported:** 16 Sep 2013 17:54

#### Performing Site Legend

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