**Lab Dept:** Anatomic Pathology  
**Test Name:** MARFAN (MFS1) SYNDROME (FBN1) TYPE I SEQUENCING  

### General Information

**Lab Order Codes:** MARF  
**Synonyms:** FBN1 Fibrillin-1 gene analysis for Marfan's syndrome; MFS1  
**CPT Codes:** 81408 – Molecular Pathology procedure, Level 9  
**Test Includes:** Testing for FBN1 gene.

### Logistics

**Test Indications:** Fibrillin-1 is the major structural component of the microfibrils that link together the various extracellular matrix components in most connective tissues, thus providing support for the organs. Microfibrils can also associate with elastin, forming elastic fibers that provide resilience and elasticity in tissues. Defects in the FBN1 gene compromise the fibrillin-1 function in these tissues and therefore, result in connective tissue weakness as seen in Marfan syndrome.

**Lab Testing Sections:** Anatomic Pathology - Sendouts  
**Referred to:** Connective Tissue Gene Tests (CTGT Test: 1187)  
**Phone Numbers:**  
- MIN Lab: 612-813-6280  
- STP Lab: 651-220-6550  
**Test Availability:** 24 hours  
**Turnaround Time:** 2 – 4 weeks  
**Special Instructions:** No transfusion within the past 30 days. Please include a completed CTGT Request form with the patient or specimen to the laboratory.

### Specimen

**Specimen Type:** Whole blood  
**Container:** Lavender top (EDTA) tube  
**Draw Volume:** 6 mL (Minimum: 3 mL) blood
**Processed Volume:** Same as Draw Volume

**Collection:** Routine blood collection, mix specimen by gentle inversion

**Special Processing:** Lab Staff: **Do Not** centrifuge. Specimen should be sent in original collection container. Send via overnight shipping with a cold pack to reach CTGT Monday through Friday. If weekend or holiday when drawn, store at refrigerated temperatures.

**Patient Preparation:** None

**Sample Rejection:** Mislabeled or unlabeled specimens

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**Interpretive**

**Reference Range:** No mutations detected

**Critical Values:** N/A

**Limitations:** The technology does not detect all possible mutations in this gene.

**Methodology:** Next Generation Sequencing (NGS)

**References:** [Connective Tissue Gene Tests] April 2018
(484) 244-2900 Fax (484) 244-2904

**Updates:**
2/6/2013: CPT update
10/29/2014: Minimum specimen volume update (previously 6 mL)
4/17/2018: TAT, volume and method update