

---

**Lab Dept:**                    **Anatomic Pathology**

**Test Name:**                **MARFAN SYNDROME (FBN1) KNOWN MUTATION**

---

***General Information***

**Lab Order Codes:**        MKM

**Synonyms:**                FBN1 Fibrillin-1 gene analysis for Marfan's syndrome

**CPT Codes:**              81403 – Molecular Pathology procedure, Level 4

**Test Includes:**            Testing for FBN1 gene.

---

***Logistics***

**Test Indications:**        This test is for family members of an affected individual who has already been tested by CTGT.

**Lab Testing Sections:**    Anatomic Pathology - Sendouts

**Referred to:**              Connective Tissue Genetics Tests (CTGT test: 1387)

**Phone Numbers:**        MIN Lab: 612-813-6280

STP Lab: 651-220-6550

**Test Availability:**        24 hours

**Turnaround Time:**        1 – 2 weeks

**Special Instructions:**    A completed [CTGT request](#) form must be sent 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. Specimens drawn on weekends or holidays should be stored at refrigerated temperatures until specimen can be shipped to arrive Monday through Friday.

**Patient Preparation:** None

**Sample Rejection:** Mislabeled or unlabeled specimens

---

***Interpretive***

**Reference Range:** No mutations detected

**Critical Values:** N/A

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

**Methodology:** Sanger sequencing

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

**Updates:** 2/6/2013: CPT update  
10/29/2014: Minimum draw vol update, previously listed as 6 mL.  
4/17/2018: TAT, volume, method update.