**Lab Dept:** Anatomic Pathology  
**Test Name:** MARFAN SYNDROME (FBN1/TGFBR2) TYPE I & II SEQUENCING

### General Information

<table>
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<tr>
<th>Lab Order Codes:</th>
<th>MAT12</th>
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<tr>
<td>Synonyms:</td>
<td>FBN1 and TGFBR2 gene analysis for Marfan’s syndrome</td>
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| CPT Codes:       | 81408 – Molecular Pathology procedure, Level 9  
81405 – Molecular Pathology procedure, Level 6 |
| Test Includes:   | Testing for genes FBN1 & TGFBR2 |

### 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 are related to Marfan Syndrome Type I, while defects in TGFBR2 are related to Marfan Syndrome Type II. |
| Lab Testing Sections: | Anatomic Pathology - Sendouts |
| Referred to: | Connective Tissue Gene Tests (CTGT Test: 1191) |
| Phone Numbers: | MIN Lab: 612-813-6280  
STP Lab: 651-220-6550 |
| Test Availability: | 24 hours |
| Turnaround Time: | 2-4 weeks |
| Special Instructions: | No transfusions within the past 30 days. Please include completed CTGT Request form with the patient or specimen to the laboratory. |

### Specimen

| Specimen Type: | Whole blood  
Additional options: Fibroblasts or Extracted DNA |
Container: Blood: Lavender top (EDTA) tube
Fibroblasts: T-25 flasks
Extracted DNA: TE solution preferred

Draw Volume: Blood: 6 mL (Minimum: 3 mL)
Fibroblasts: 4 confluent T-25 flasks
Extracted DNA: a minimum of 30 mcgrams of genomic DNA at a concentration of 100 ng/mcL or more, preferably in TE solution

Processed Volume: Same as Draw Volume

Collection: Routine blood collection, mix specimen by gentle inversion

Special Processing: Lab Staff: Blood: Do Not centrifuge.
Blood and other specimens 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

Interpretive

Reference Range: No mutations detected

Critical Values: N/A

Limitations: The technology does not detect all possible mutations in these genes.

Methodology: Next Generation Sequencing

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

Update: 2/6/2013: CPT update
7/10/2013: CPT update CTGT, now reported as both 81408 & 81405
10/29/2014: Additional specimen types added. Blood min updated, previously listed as 6 mL.