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

**Test Name:**                **MARFAN SYNDROME (TGFB2) TYPE II SEQUENCING**

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***General Information***

**Lab Order Codes:**        MARF2

**Synonyms:**                TGFB2 - gene analysis for Marfan's syndrome  
TGFB2 sequencing also applies to Loeys Dietz Type 1B and Aortic Aneurysm, Familial Thoracic Aortic Syndromes.

**CPT Codes:**                81405 – Molecular Pathology procedure, Level 6

**Test Includes:**            Sequencing for the TGFB2 gene.

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***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 TGFB2 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)

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

STP Lab: 651-220-6550

**Test Availability:**        24 hours

**Turnaround Time:**        2 to 3 months

**Special Instructions:**    No transfusion within the past 30 days. Please include a completed CTGT [Request Form](#) with the patient or specimen to the laboratory.

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***Specimen***

**Specimen Type:**            Whole blood

**Container:**                 Lavender top (EDTA) tube

<b>Draw Volume:</b>	6 mL (Minimum: 3 mL) blood
<b>Processed Volume:</b>	Same as Draw Volume
<b>Collection:</b>	Routine venipuncture, mix specimen by gentle inversion
<b>Special Processing:</b>	Lab Staff: <b>Do Not</b> centrifuge. Specimen should be sent in original collection container. Send via overnight shipping to reach CTGT Monday through Friday. If weekend or holiday when drawn, store at refrigerated temperatures.
<b>Patient Preparation:</b>	None
<b>Sample Rejection:</b>	Mislabeled or unlabeled specimens

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### ***Interpretive***

<b>Reference Range:</b>	No mutations detected
<b>Critical Values:</b>	N/A
<b>Limitations:</b>	The technology does not detect all possible mutations in this gene.
<b>Methodology:</b>	PCR amplification, gel electrophoresis
<b>References:</b>	<a href="#">Connective Tissue Gene Tests</a> September 2010 (484) 224-2900 Fax (484) 244-2904
<b>Updates:</b>	2/11/2013: CPT update