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)
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.

Specimen

Specimen Type:	Whole blood
Container:	Lavender top (EDTA) tube
Draw Volume:	9 mL (Minimum: 4 mL) blood

Processed Volume:	Same as Draw Volume
Collection:	Routine venipuncture, 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

Interpretive

Reference Range:	No mutations detected
Critical Values:	N/A
Limitations:	The technology does not detect all possible mutations in this gene.
Methodology:	PCR amplification, gel electrophoresis
References:	Connective Tissue Gene Tests October 2014 (484) 244-2900 Fax (484) 244-2904
Updates:	2/6/2013: CPT update 10/29/2014: Minimum specimen volume update (previously 6 mL)