
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

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