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

General Information

Lab Order Codes:	MAT12
Synonyms:	FBN1 and TGFBR2 gene analysis for Marfan's syndrome
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. 4/17/2018: Volume, TAT and method update.