Lab Dept: Anatomic Pathology

Test Name: MARFAN SYNDROME (FBN1) KNOWN MUTATION

General Information

Lab Order Codes:	MKM
Synonyms:	FBN1 Fibrillin-1 gene analysis for Marfan's syndrome
CPT Codes:	81403 – Molecular Pathology procedure, Level 4
Test Includes:	Testing for FBN1 gene.

Logistics

Test Indications:	This test is for family members of an affected individual who has already been tested by CTGT.
Lab Testing Sections:	Anatomic Pathology - Sendouts
Referred to:	Connective Tissue Genetics Tests (CTGT test: 1387)
Phone Numbers:	MIN Lab: 612-813-6280 STP Lab: 651-220-6550
Test Availability:	24 hours
Turnaround Time:	1 – 2 weeks
Special Instructions:	A completed CTGT request form must be sent 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. Specimens drawn on weekends or holidays should be stored at refrigerated temperatures until specimen can be shipped to arrive Monday through Friday.
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:	Sanger sequencing
References:	Connective Tissue Gene Tests April 2018 (484) 244-2900 Fax (484) 244-2904
Updates:	2/6/2013: CPT update 10/29/2014: Minimum draw vol update, previously listed as 6 mL. 4/17/2018: TAT, volume, method update.