Marfan Syndrome (FBN1)

Order Name: **FBN1 Marfan Synd**
Test Number: 5194960
Revision Date: 03/21/2023

TEST NAME	METHODOLOGY	LOINC CODE
Marfan Syndrome (FBN1)	Polymerase Chain Reaction	

SPECIMEN REQUIREMENTS

Specimen	Specimen Volume (min)	Specimen Type	Specimen Container	Transport Environment
Preferred	**8.5 mL (3 mL)**	**Whole Blood**	**ACD Solution A or B (Yellow Top)**	**Room Temperature**
Alternate 1	**8.5 mL (3 mL)**	**Whole Blood**	**EDTA (Lavender Top)**	**Room Temperature**
Alternate 2	**1**	**Saliva**	**Oragene Dx saliva kit**	**Room Temperature**
Alternate 3	**1**	**Buccal swab**	**PurFlock buccal swab kit**	**Room Temperature**

Instructions

Specimen Type: Whole blood or PurFlock buccal swab kit or Oragene Dx saliva kit
Specimen Volume: 8.5 mL whole blood or PurFlock buccal swab kit or Oragene Dx saliva kit
Mininum Volume: 3 mL whole blood or PurFlock buccal swab kit or Oragene Dx saliva kit
Collection: Standard phlebotomy. Follow PurFlock buccal swab kit or Oragene Dx 500 saliva kit collection instructions. Do not eat, drink, smoke, or chew gum 30 min prior to collection.
Specimen Storage: Maintain specimen at room temperature or refrigerate at 4C Do not freeze.
Special Instructions: In cases in which there is a known variant documented in the family, the physician may prefer to order **Targeted Variant Analysis**, test code **5194970**. Test orders must include an attestation that the provider has the patient's informed consent for genetic testing.

GENERAL INFORMATION

Expected TAT	14 - 21 days In some cases, additional time may be required for confirmatory or reflex tests.
Clinical Use	This test is used for diagnostic testing for Marfan syndrome and presymptomatic testing for family members. Next-generation sequencing: Identifies genetic variants, including small nucleotide variants (SNVs), insertions, deletions and copy number variants (CNVs).
Performing Labcorp Test Code	482336
Notes	Labcorp Test Code: 482336
Lab Section	Reference Lab