

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	<p><b>Specimen Type:</b> Whole blood or PurFlock buccal swab kit or Oragene Dx saliva kit</p> <p><b>Specimen Volume:</b> 8.5 mL whole blood or PurFlock buccal swab kit or Oragene Dx saliva kit</p> <p><b>Mininum Volume:</b> 3 mL whole blood or PurFlock buccal swab kit or Oragene Dx saliva kit</p> <p><b>Collection:</b> 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.</p> <p><b>Specimen Storage:</b> Maintain specimen at room temperature or refrigerate at 4C Do not freeze.</p> <p><b>Special Instructions:</b> In cases in which there is a known variant documented in the family, the physician may prefer to order <b>Targeted Variant Analysis</b>, test code <b>5194970</b>. Test orders must include an attestation that the provider has the patient's informed consent for genetic testing.</p>			

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