
Reporting Title: FBN1 Full Gene Analysis**Performing Location:** Rochester**Ordering Guidance:**

This is a single gene test for the FBN1 gene. The FBN1 gene is also included on multi-gene panels. If testing for multiple overlapping clinical presentations is desired, see MFRGG Marfan, Loeys-Dietz, and Aortopathy Gene Panel, Varies or CAORG / Comprehensive Marfan, Loeys-Dietz, Ehlers-Danlos, and Aortopathy Gene Panel, Varies.

Customization of this panel and single gene analysis for any gene present on this panel are available. For more information see CGPH / Custom Gene Panel, Hereditary, Next-Generation Sequencing, Varies.

Targeted testing for familial variants (also called site-specific or known mutations testing) is available for the genes on this panel. See FMTT / Familial Variant, Targeted Testing, Varies. To obtain more information about this testing option, call 800-533-1710.

Shipping Instructions:

Specimen preferred to arrive within 96 hours of collection.

Necessary Information:

Prior Authorization is available, but not required, for this test. If proceeding with the prior authorization process, submit the required form with the specimen.

Specimen Requirements:

Patient Preparation: A previous bone marrow transplant from an allogenic donor will interfere with testing. Call 800-533-1710 for instructions for testing patients who have received a bone marrow transplant.

Specimen Type: Whole blood

Container/Tube:

Preferred: Lavender top (EDTA) or yellow top (ACD)

Acceptable: Any anticoagulant

Specimen Volume: 3 mL

Collection Instructions:

1. Invert several times to mix blood.
2. Send whole blood specimen in original tube. Do not aliquot.

Specimen Stability Information: Ambient (preferred)/Refrigerated

Specimen Minimum Volume:

1 mL

Forms:

1. New York Clients-Informed consent is required. Document on the request form or electronic order that a copy is on file. The following documents are available:
- Informed Consent for Genetic Testing (T576)
 - Informed Consent for Genetic Testing (Spanish) (T826)
2. Connective Tissue/Cerebrovascular Disease Genetic Testing Patient Information
3. FBN1 Full Gene Analysis (MFBNG) Prior Authorization Ordering Instructions
4. If not ordering electronically, complete, print, and send a Cardiovascular Test Request Form (T724) with the specimen.

Specimen Type	Temperature	Time	Special Container
Varies	Varies		

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
617366	Test Description	Alphanumeric		62364-5
617367	Specimen	Alphanumeric		31208-2
617368	Source	Alphanumeric		31208-2
617369	Result Summary	Alphanumeric		50397-9
617370	Result	Alphanumeric		82939-0
617371	Interpretation	Alphanumeric		69047-9
617372	Additional Results	Alphanumeric		82939-0
617373	Resources	Alphanumeric		99622-3
617374	Additional Information	Alphanumeric		48767-8
617375	Method	Alphanumeric		85069-3
617376	Genes Analyzed	Alphanumeric		48018-6
617377	Disclaimer	Alphanumeric		62364-5
617378	Released By	Alphanumeric		18771-6

LOINC and CPT codes are provided by the performing laboratory.

Supplemental Report:
Supplemental**CPT Code Information:**

81408

Reference Values:

An interpretive report will be provided.