

Using Genetic Testing for Timely Diagnosis of Marfan Syndrome (MFS)

Indications:

- Presence of at least one major and one minor criterion according to the Ghent criteria for diagnosis of MFS
- Confirmed family history of MFS or another type I fibrillinopathy

Benefits:

Genetic testing for MFS-related *FBN1* mutations can:

- confirm a clinical diagnosis of MFS.¹
- allow a diagnosis of MFS in patients with clinically “incomplete” MFS according to the Ghent criteria.¹
- indicate risk of aortic dilation in other type I fibrillinopathies such as familial ectopia lentis, to guide treatment choice.²
- exclude increased risk of MFS in presymptomatic family members of index patients with known *FBN1* mutations.³

Background:

- Marfan syndrome (MFS) is a systemic connective tissue disorder that is characterized by multiple variable abnormalities of the skeletal, ocular, cardiovascular, pulmonary, skin, and nervous systems.⁴
- MFS occurs at a prevalence of 1 in 5,000 with an autosomal dominant mode of inheritance and, in most cases, is caused by mutations in the gene *FBN1*.⁴
- MFS is associated with aortic dilation leading to aortic dissection or rupture.⁴
- Timely initiation of drug therapy can slow aortic dilation, and timely elective aortic root replacement can prevent aortic dissection.⁵
- Early diagnosis of MFS is complicated by variable expression of the disease and the gradual onset of characteristic manifestations.
- Presence of a pathogenic *FBN1* mutation can indicate risk of aortic dilation even in the absence of a clinically diagnostic combination of manifestations.^{1,2}

References: 1. Faivre L, et al (2008) J Med Genet epub ahead of print 2. Pepe G, et al (2007) Molecular Vision 13:2242. 3. Comeglio P, et al (2007) Hum Mutat 28:928. 4. Judge DP & Dietz HC (2005) Lancet 366:1965. 5. Judge DP & Dietz HC (2008) Annu Rev Med 59:43.

Ordering Information: please see other side

Ordering Information for Marfan Syndrome (MFS) Testing

Indications for Testing

- Presence of at least one major and one minor criterion according to the Ghent criteria for diagnosis of MFS
- Confirmed family history of MFS or another type I fibrillinopathy

Ordering Information

Gene	CPT Codes	Test Code
FBN1	83891(1) 83892(1) 83898(62) 83904(124) 83909(124) 83912(1)	190601

Ordering Information for Multi-Gene Panels

FBN1, TGFB1, TGFB2	83891(1) 83892(1) 83898(80) 83904(160) 83909(160) 83912(1)	190699
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Reflexive testing option

FBN1 tested first → if no clear or probable disease variant is found, reflexing to **TGFB1** and **TGFB2** testing 190650

TGFB1, TGFB2	83891(1) 83892(1) 83898(18) 83904(36) 83909(36) 83912(3)
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Family Testing (single amplicon)

FBN1	83891(1) 83892(1) 83898(1) 83904(2) 83909(2) 83912(1)	190601
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Test Methodology

- Amplification by polymerase chain reaction (PCR); sequencing of entire protein-coding region as well as splice donor and acceptor sites

Sample Requirements

- 2 mL whole blood in EDTA tube (lavender top)
- Samples can be stored briefly at 4°C, but should be shipped on day of collection
- All sample types should be shipped overnight at room temperature
- To request a sample shipping kit, please call 1-866-647-0735

**For more information, please contact us at 1-866-647-0735
or visit us on the web at www.correlagen.com.**