

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	Test Code
<i>FBN1</i>	190601

Ordering Information for Multi-Gene Panels

<i>FBN1, TGFB1, TGFB2</i>	190699
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Reflexive testing option

<i>FBN1</i> tested first → if no clear or probable disease variant is found, reflexing to <i>TGFB1</i> and <i>TGFB2</i> testing	190650
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Family Testing (single amplicon)

Family Testing is available. Please contact Client Services at 1-866-647-0735 for requirements.

Test Methodology

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

NOTE: Specimens must be accompanied by a completed consent form. In the case of family tests (ie, known mutations), a copy of the result of the first patient tested in the family (the index case) must be submitted unless that test was performed at Correlagen. Other family members are subsequently tested for the specific mutation found in the first patient tested.

For test information, sample requirements, or to request a sample shipping kit, please contact Client Services at 1-866-647-0735 or visit us on the web at www.correlagen.com.