

# Genetic Testing for Marfan Syndrome (MFS)

## What is Marfan Syndrome?

*Marfan Syndrome is an inherited disease that can lead to rupture of the body's main blood vessel.*

Marfan Syndrome (MFS) is an inherited connective tissue disorder that affects many different body systems, including bones, eyes, skin, and the cardiovascular (heart and blood vessels) system. Individuals with MFS are typically tall and thin for their family background, with long arms and legs and long fingers and toes, and have bad eye sight. The dangerous consequences of MFS, however, are less obvious from the outside. MFS often leads to stretching (dilation) of the aorta, the main blood vessel carrying blood from the heart to the rest of the body. If left untreated, the over-stretched aorta will eventually split (dissect) or rupture, leading to death. Luckily, this devastating outcome can be prevented through timely treatment and preventive surgery.

## How is Marfan Syndrome treated?

*Marfan Syndrome is treated with medication and preventive surgery.*

Aortic dilation can be slowed through drug treatment, and aortic dissection or rupture can be prevented by surgically replacing the dilated part of the aorta before it ruptures. In addition, individuals with MFS can make some precautionary lifestyle adjustments. They should avoid contact sports, competitive sports, and isometric exercise, which increase the risk of aortic rupture. In some individuals with MFS, activities that involve breathing against resistance, such as scuba diving or playing a wind instrument, are also not advised since these can promote episodes of spontaneous lung collapse (pneumothorax).

## What causes Marfan Syndrome?

*Marfan Syndrome is caused by variations in the gene *FBNI*.*

Most or all cases of MFS are due to a genetic defect in the gene *FBNI*. If an individual harbors an MFS-associated genetic defect in *FBNI*, each of his or her children has a 50% chance of inheriting this defect, and thus, the condition.

## How is Marfan Syndrome diagnosed?

*Marfan Syndrome is usually diagnosed based on the presence of a characteristic combination of several different symptoms.*

Characteristic signs of Marfan Syndrome are often common by themselves and indicate Marfan Syndrome only if they occur in combination. They include taller stature than expected from family background, a slender body build, very long arms and legs, very long fingers and toes, a protruding or a sunken chest, near-sightedness with astigmatism, overly flexible joints, and stretch marks on the skin in the absence of weight gain. In addition, there are other, less obvious characteristic signs that a physician can detect.

The so-called "Ghent criteria" define how many and which of these symptoms have to be present before a diagnosis of MFS can be reached. (The Ghent criteria are named for the Belgian town of Ghent, where a panel of experts convened to define these diagnostic criteria.)



## How can genetic testing help individuals and families with Marfan Syndrome?

*Genetic testing can help to diagnose MFS in individuals before a characteristic combination of symptoms has appeared.*

*If the familial mutation is known, genetic testing can help to identify family members who are unlikely to develop MFS and do not need treatment or monitoring.*

Diagnosis of MFS based on symptoms is complicated by the fact that (1) many of the symptoms by themselves are relatively common, (2) some symptoms only appear over time, and (3) some of the characteristic symptoms may be severe in one individual with MFS and mild in another. Genetic testing can help to achieve a diagnosis of MFS before a characteristic combination of symptoms has developed, facilitating timely initiation of drug treatment and regular monitoring of the degree of aortic dilation. If the specific mutation affecting an extended family (the familial mutation) is known, genetic testing may also allow identification of family members who are not at risk of MFS, even if they exhibit some of the more common symptoms of MFS.

## If you still have questions about Marfan Syndrome, please contact us or consult any of the other resources listed below:

### Correlagen Diagnostics

[www.correlagen.com](http://www.correlagen.com)  
phone: 1-866-647-0735  
email: [testing@correlagen.com](mailto:testing@correlagen.com)

### National Marfan Foundation (NMF)

[www.marfan.org](http://www.marfan.org)  
phone: 1-800-862-7326  
email: [staff@marfan.org](mailto:staff@marfan.org)

### National Heart Lung and Blood Institute

[http://www.nhlbi.nih.gov/health/dci/Diseases/mar/mar\\_what.html](http://www.nhlbi.nih.gov/health/dci/Diseases/mar/mar_what.html)

### National Institute of Arthritis and Musculoskeletal and Skin Diseases

[http://www.niams.nih.gov/Health\\_Info/Marfan\\_Syndrome/default.asp](http://www.niams.nih.gov/Health_Info/Marfan_Syndrome/default.asp)

### Additional reading:

1. Judge DP, Dietz HC (2005) Marfan's syndrome. *Lancet* 366:1965-1976.
2. Dietz HC (2005) Marfan syndrome. *Gene Reviews*. [www.genetests.org](http://www.genetests.org).
3. Judge DP, Dietz HC (2008) Therapy of Marfan Syndrome. *Annu Rev Med* 59:43-59.



[www.correlagen.com](http://www.correlagen.com)  
Copyright © 200  
Correlagen Diagnostics, Inc.  
All rights reserved  
MFS Patient Info 05/08