

Facts on Genetic Testing for Marfan Syndrome (MFS)

- **Marfan syndrome (MFS) is a dominantly inherited connective tissue disorder that affects 1 in 5,000 individuals. A mutation in the gene *FBN1* can be detected in about 90% of patients with a clinical diagnosis of MFS according to the Ghent criteria.**
 - [Judge DP, Dietz HC \(2005\) Lancet 366:1965.](#)
- **MFS is associated with a high risk of aortic dilation that, if left untreated, usually leads to fatal aortic dissection or rupture before the age of 40.**
 - [Murdoch JL et al \(1972\) N Engl J Med 286: 804.](#)
- **MFS-related aortic dilation can be slowed through timely pharmacologic treatment.**
 - [Judge DP, Dietz HC \(2008\) Annu Rev Med 59:43.](#)
- **Aortic dissection or rupture can be prevented through elective aortic root replacement in patients with high degrees or rates of aortic dilation.**
 - [Gott VL, et al. \(1999\) N. Engl. J. Med. 340:1307](#)
- **Timely pharmacologic treatment and effective monitoring of aortic dilation depend on early diagnosis.**
- **Early diagnosis of MFS is complicated by the gradual onset and the variable nature and expression of manifestations that form the basis for clinical diagnosis according to the Ghent criteria**
 - [Faivre L, et al \(2008\) J Med Genet epub ahead of print](#)
- **Genetic testing for pathogenic mutations in *FBN1* can confirm a diagnosis of MFS and indicate risk of aortic dilation before a characteristic combination of clinical manifestations has developed.**

In a large series of probands, genetic testing allowed diagnosis of MFS in about half of adults with “incomplete” MFS according to clinical criteria alone.

 - [Faivre L, et al \(2008\) J Med Genet epub ahead of print](#)
- **In presymptomatic relatives of probands with known *FBN1* mutations, genetic testing can exclude risk by showing absence of the familial mutation.**

Genetic testing could exclude increased risk of aortic dilation in 32 out of 51 presymptomatic young relatives of MFS patients with known *FBN1* mutations.

 - [Comeglio et al \(2007\) Hum Mutat 28:928](#)