



How to Find Out if You Have Marfan Syndrome

People sometimes ask, "Do I or a loved one have Marfan syndrome?" Here are some ways you and your doctor can find out the answer to this question.

WHAT YOU CAN DO

Find a doctor who knows about Marfan syndrome (MFS)

Marfan syndrome (MFS) is rare and not all doctors know about it. You need to find a doctor who knows about MFS and how to tell the difference between MFS and the other medical conditions that look like it.

The first choice of doctor to look for is a medical geneticist (a doctor who specializes in genetic conditions such as Marfan syndrome).

A second choice is a cardiologist (heart doctor). Make sure the cardiologist has treated people who have MFS.

You can find a doctor by:

- asking your primary doctor for a referral
- calling the doctor referral service at your local hospital
- calling the National Marfan Foundation Resource Center at 800-862-7326, ext. 26

Learn the health history of you and your family. You might want to write this down in a health history notebook with lists of:

- your past illnesses, operations, and hospitalizations
- medications you are taking
- reasons why you think you might have MFS
- family members who have, or might have, MFS
- family members who died of a heart problem



National Marfan Foundation

Education • Research • Support

Serving the needs
of people with
Marfan syndrome
and related disorders

22 Manhasset Avenue
Port Washington, NY 11050

516-883-8712
800-8-MARFAN
516-883-8040 (fax)

www.marfan.org

WHAT YOUR DOCTOR CAN DO

Talk with you about your health history. This is the time when you can talk about the information in your health history notebook and why you think you might have MFS.

Do a thorough physical exam. This includes looking for MFS features in your bones, joints, skin, and lungs.

Ask you to have medical tests. These common, painless tests include:

- Echocardiogram. This test looks at the heart, its valves, and the aorta (vessel that carries blood from the heart).
- Electrocardiogram (EKG). This test checks your heart rate and heart rhythm. Your doctor may want to do an EKG in addition to an echocardiogram.
- Slit lamp eye exam. This test, a part of most eye exams, helps your doctor see if the lenses in your eyes are dislocated (out of place).
- Other tests, such as a MRI or CT scan of the lower back. These tests can help your doctor see if you have dural ectasia, a back problem that is very common in people who have MFS.

WHAT YOU AND YOUR DOCTOR CAN DO

You asked, “Do I have MFS?” There are three possible answers to this question. Each answer has actions you and your doctor should take.

- **No, you do not have MFS.** Ask your doctor if you have any other medical condition that needs medical care. You should also ask how you can get this needed care.
- **Maybe, you have MFS.** Ask your doctor if you should repeat the echocardiogram in 1 to 2 years to find out if the valves of your heart and aorta have changed.
- **Yes, you have MFS.** Ask your doctor how to take care of yourself. It is very important to follow these instructions. Find out if other people in your family also have MFS. You may feel overwhelmed and have a lot of questions. Below are some ways to learn more about MFS.

WAYS TO LEARN MORE ABOUT MFS

- Call the National Marfan Foundation (NMF) Resource Center at 1-800-862-7326 ext. 26. You will speak with a nurse who can answer your questions and mail you information. She can also suggest articles your doctor can read about MFS.
- Talk with your doctor. Sometimes it helps to use the information you have from the NMF when you speak with your doctor.
- Visit the NMF website at <http://www.marfan.org>. You can print out information from the “About Marfan Syndrome” and “Living with Marfan Syndrome” pages. You can also ask questions online by clicking: “Support Services: Ask a Question.”