



How to Get Diagnosed with Marfan Syndrome

Marfan syndrome is a serious, potentially life-threatening disorder, and an early, accurate diagnosis is essential, not only for people with Marfan syndrome, but also for those with related disorders.

How is Marfan syndrome diagnosed?

A Marfan diagnosis can often be made after exams of several parts of the body by doctors experienced with connective tissue disorders, including:

- A detailed medical and family history, including information about any family member who may have the disorder or who had an early, unexplained, heart-related death. A complete physical examination.

You should also have tests to identify Marfan features that are not visible during the physical exam, including:

- Echocardiogram. This test looks at your heart, its valves, and the aorta (blood vessel that carries blood from the heart to the rest of the body).
- Electrocardiogram (EKG). This test checks your heart rate and heart rhythm.
- An eye examination, including a “slit lamp” evaluation to see if the lenses in your eyes are out of place. It is important that the doctor fully dilates the pupils.
- Other tests, such as a computerized tomography (CT) scan or magnetic resonance imaging (MRI) of the lower back. These tests can help your doctor see if you have a back problem that is very common in people with Marfan syndrome called dural ectasia (swelling of the sac around the spinal column).

Is there a genetic test for Marfan syndrome?

Genetic testing can provide helpful information in some cases.

- For individuals with a family history of Marfan syndrome, genetic testing can help confirm or rule out the diagnosis of Marfan syndrome in family members who may be at risk.
- Some of the features of Marfan syndrome can be found in disorders related to Marfan syndrome; therefore, genetic testing may be helpful when a diagnosis cannot be determined through an exam by doctors.

What if you have some features of Marfan syndrome, but the doctor says you don't have Marfan syndrome?

It is possible for you to have one or more features of Marfan syndrome, but not enough for you to have a Marfan syndrome diagnosis. You may need additional exams by other doctors and additional genetic testing to see if you have a disorder that is related to Marfan syndrome. Read about the disorders related to Marfan syndrome and how each is diagnosed.

What should you do if you think you might have Marfan syndrome or a related disorder?

If you suspect that you or a family member may have Marfan syndrome or a related disorder, we recommend that you:

- Find a doctor who is knowledgeable about genetic disorders
- Compile a health history
- Reach out for information and support

Contact The Marfan Foundation to learn more. Marfan.org or 800-8-MARFAN.