



Marfan Syndrome: Diagnostic Criteria

Marfan Syndrome Diagnostic Criteria is a list of features doctors use to diagnose (decide if someone has) Marfan syndrome. Some of these features are easy to see. Others need special tests to find them. Diagnostic criteria are sometimes called “Ghent Criteria,” named after the city in Belgium where doctors decided which features to include on the list.

Here are some facts about Marfan syndrome diagnostic criteria.

MAJOR AND MINOR DIAGNOSTIC CRITERIA

There are both major and minor diagnostic criteria. “Major criteria” refers to features that are common in people with Marfan syndrome, but rare in others. “Minor criteria” are features that are common in many people—those with and without Marfan syndrome.

Since Marfan syndrome affects many body systems, there are diagnostic criteria for many parts of the body. You can find a detailed list of these criteria in the fact sheet, “Marfan Syndrome Diagnostic Criteria by Body System.”

HOW DOCTORS USE THE DIAGNOSTIC CRITERIA

To diagnose Marfan syndrome, doctors compare a person’s test results, health history, and physical examination with the diagnostic criteria.

- If no one else in the family has Marfan syndrome, then doctors diagnose Marfan syndrome when a person has major criteria in at least two body systems and a minor criteria in a third.
- If a parent or sibling (brother or sister) has Marfan syndrome, then doctors diagnose Marfan syndrome when a person has a major criteria in one body system and a minor criteria in another.

Doctors use diagnostic criteria to diagnose Marfan syndrome because the genetic (DNA) test for Marfan syndrome does not always show if a person has the disorder.

It can be hard for even skilled doctors to make this diagnosis as features may be due to Marfan syndrome or happen for other reasons. Doctors who see many people with Marfan syndrome are more likely to make the correct diagnosis.



National Marfan Foundation

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and related disorders

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TYPES OF DIAGNOSES

A person with Marfan features may or may not have Marfan syndrome. Here are some diagnoses that doctors can make:

- **Marfan syndrome.** A doctor makes this diagnosis when a person has the major and minor criteria as described above. In these cases, it can be said that a person “meets” the diagnostic criteria for Marfan syndrome.
- **Some other genetic disorder of connective tissue.** A person who does not meet the diagnostic criteria for Marfan syndrome may instead have another genetic disorder of connective tissue. These disorders include: Ehlers-Danlos syndrome, Loeys-Dietz syndrome, MASS phenotype, familial aortic aneurysm, and Sticklers syndrome.
- **Emerging Marfan syndrome.** Doctors may use this term when a child has just some of the diagnostic criteria for Marfan syndrome. This is likely when doctors think the child will have more features as the child grows older.
- **No specific diagnosis.** This term is used when a person has Marfan features but does not meet the diagnostic criteria for any known disorder.

It is important to understand that a person can have many Marfan features but not have Marfan syndrome. For instance, a person may have many skeletal features but that would only count as one major criteria—that being of the skeletal system. The same person would need to have criteria in other body systems in order to be diagnosed with Marfan syndrome.

TREATMENT AND FOLLOW-UP CARE

Many people with Marfan features (whether they have a diagnosis or not) need medical treatment and follow-up care. Make sure to talk with your doctor about the care that is right for you.

WAYS TO LEARN MORE:

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