THE MARFAN FOUNDATION

Know the signs. Fight for victory.

What is Marfan Syndrome?

Marfan syndrome is a genetic disorder that affects the body's connective tissue. Connective tissue holds all the body's cells, organs and tissue together. It also plays an important role in helping the body grow and develop properly.

Connective tissue is made up of proteins. The protein that plays a role in Marfan syndrome is called fibrillin-1. Marfan syndrome is caused by a defect (or mutation) in the gene that tells the body how to make fibrillin-1. This mutation results in an increase in a protein called transforming growth factor beta, or TGF-β. The increase in TGF-β causes problems in connective tissues throughout the body, which in turn creates the features and medical problems associated with Marfan syndrome and some related disorders.

Because connective tissue is found throughout the body, Marfan syndrome can affect many different parts of the body, as well. Features of the disorder are most often found in the heart, blood vessels, bones, joints, and eyes. Some Marfan features – for example, aortic enlargement (expansion of the main blood vessel that carries blood away from the heart to the rest of the body) – can be life-threatening. The lungs, skin and nervous system may also be affected. Marfan syndrome does not affect intelligence.

Who has Marfan syndrome?

About 1 in 5,000 people have Marfan syndrome, including men and women of all races and ethnic groups. About 3 out of 4 people with Marfan syndrome inherit it, meaning they get the genetic mutation from a parent who has it. But some people with Marfan syndrome are the first in their family to have it; when this happens it is called a spontaneous mutation. There is a 50 percent chance that a person with Marfan syndrome will pass along the genetic mutation each time they have a child.

Knowing the signs of Marfan syndrome can save lives

People with Marfan syndrome are born with it, but features of the disorder are not always present right away. Some people have a lot of Marfan features at birth or as young children – including serious conditions like aortic enlargement. Others have fewer features when they are young and don't develop aortic enlargement or other signs of Marfan syndrome until they are adults. Some features of Marfan syndrome, like those affecting the heart and blood vessels, bones or joints, can get worse over time.

This makes it very important for people with Marfan syndrome and related disorders to receive accurate, early diagnosis and treatment. Without it, they can be at risk for potentially life-threatening complications. The earlier some treatments are started, the better the outcomes are likely to be.

Knowing the signs of Marfan syndrome can save lives. Our community of experts estimates that nearly half the people who have Marfan syndrome don't know it. This is something we are working hard to change.

Learn more about the signs of Marfan syndrome at Marfan.org.

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