Marfan Syndrome

Introduction

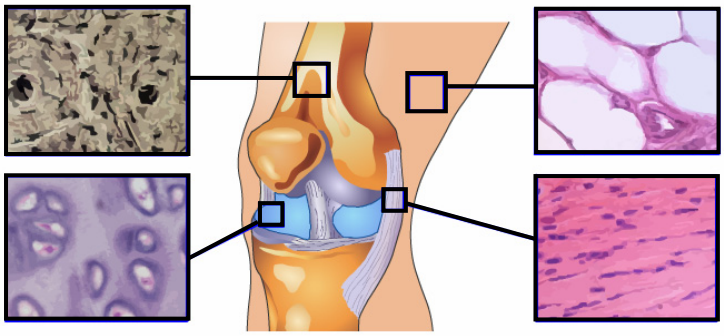
Marfan syndrome is a condition in which your body's connective tissue is abnormal. Connective tissue helps support all parts of your body. It also helps control how your body grows and develops. Most people with Marfan syndrome have heart and blood vessel problems. They may also have problems with their bones, eyes, skin, nervous system and lungs.

This reference summary explains Marfan syndrome. It covers symptoms, causes, diagnosis and treatment.

What is Marfan Syndrome?

Connective tissue is the material inside your body that supports many of its parts. It is the "cellular glue" that gives your tissues their shape and helps keep them strong. It also helps some of your tissues do their work. Cartilage and fat are examples of connective tissue.

Marfan syndrome is a genetic disease that causes a defect in the gene that produces the protein fibrillin. Fibrillin is a protein that plays a major role in your body's connective tissue.



Connective Tissue

Connective tissues provide strength and flexibility to structures in the body, such as bones, muscles, and blood vessels.

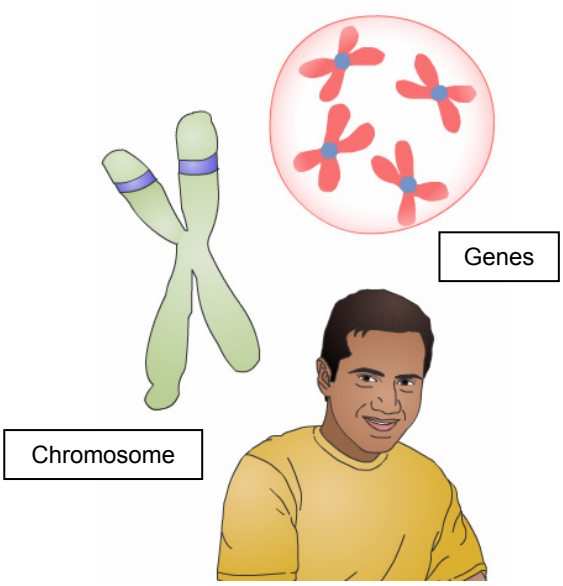
Problems with fibrillin cause tissues in the body to stretch and become weak. Not having enough fibrillin can also affect the growth and repair of tissues throughout the body.

Last reviewed: 12/18/2018

Causes

Marfan syndrome is a genetic disorder. This means that a mutation, or change, in a specific gene causes Marfan syndrome.

Genes tell the body to make certain substances. Every person has thousands of genes. Our genes make us look the way we do. They also have a lot to do with our health.



Each person has 23 pairs of chromosomes. Genes are found on chromosomes. The gene linked to Marfan syndrome is found on chromosome 15. It is called the FBN1 gene.

The FBN1 gene makes a protein called fibrillin. Fibrillin is one of many proteins that form connective tissue in the body.

The FBN1 gene either works correctly or it is defective. We will call the good FBN1 gene that works correctly “g” for “good.” We will call the defective FBN1 gene that does not work correctly “D” for “defective.”

Often Marfan syndrome is inherited. This means that th defective gene that causes Marfan syndrome is passed on from parent to child.

Everybody has two FBN1 genes: 1 from their father and 1 from their mother. If one FBN1 gene is defective, the person will have Marfan syndrome. Each parent has two copies of every chromosome but only gives one to each child. Which gene a child gets from each parent is random.

If both parents are gg, their child will not have the defective gene that causes Marfan syndrome. Each parent can only give one good FBN1 gene. If one parent is gD, meaning they have the defective FBN1 gene, their child has a 50-50 chance of getting it. The parent may pass on the good “g” gene or they may pass on the defective “D” gene.

If a child does not inherit the defective FBN1 gene, he or she will not develop Marfan syndrome and cannot pass it on to their children. A person that inherits the defective gene will have Marfan syndrome and can pass it on to their children.

Last reviewed: 12/18/2018

In some families with an affected parent, all children may inherit the defective FBN1 gene. In other families, none do. If one child has the defective gene, it does not mean that all other children born into the family will inherit it.

Less often, some people may develop Marfan syndrome without having a family history of it. This is due to a new genetic change in the FBN1 gene that happens in the egg or sperm cells.

Symptoms and Complications

Marfan syndrome can be mild to severe and can affect many parts of the body. Symptoms of Marfan syndrome vary from person to person, even in the same family.

Marfan syndrome often affects the long bones of the body. This leads to the following traits:

- A tall, thin body frame
- Flat feet
- Flexible joints
- Long arms, legs, fingers, and toes
- Teeth that are too crowded

Another common symptom of Marfan syndrome is a chest that sinks in or sticks out. If the chest sinks in, it is called pectus excavatum. Pectus carinatum is the name for a chest that sticks out. The spine may also curve to one side, which is called scoliosis.

Stretch marks on the skin are also a common trait of people who have Marfan syndrome. Stretch marks usually appear on the:

- Abdomen
- Breasts
- Buttocks
- Lower back
- Shoulders
- Thighs

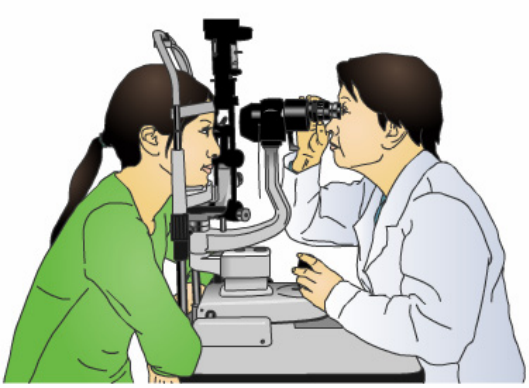
Last reviewed: 12/18/2018

Most people with Marfan syndrome have serious problems with the heart and blood vessels. This may include:

- A weak part of the aorta, which is the main blood vessel that supplies oxygen-rich blood to the body
- A torn or ruptured aorta
- Heart valves that leak, causing a heart murmur, which is an extra or unusual sound hear during the heartbeat

Some people with Marfan syndrome have problems with the eyes, which can affect eyesight. These problems can include:

- A detached retina in the eye
- A shift in one or both lenses of the eye
- Glaucoma, which is high pressure in the fluid in the eyes, at a young age
- Cataracts, which are clouding of an eye's lens, at a young age
- Nearsightedness



When people with Marfan syndrome get older, they may have problems with the connective tissue covering the brain and spinal cord. The connective tissue may weaken and stretch. This affects the bones in the lower spine, causing painful, numb or weak legs.

People with Marfan syndrome sometimes have lung problems. Symptoms may include:

- Stiff air sacs in the lungs, which may make breathing difficult
- A collapsed lung
- Sleep apnea, which is a disorder in which breathing pauses or gets very shallow when sleeping

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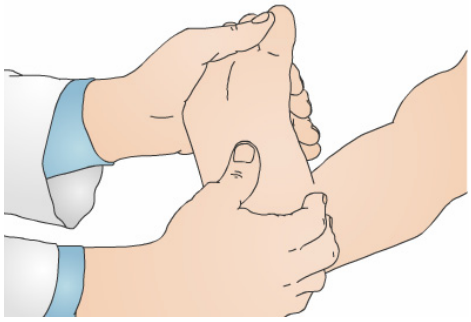
Diagnosis

To diagnose Marfan syndrome, your healthcare provider will ask about your symptoms and your family medical history.



Your healthcare provider will also perform a physical exam to check for common traits of Marfan syndrome. This includes measuring the length of bones in the arms and legs. He or she may also check the curve of your spine or shape of your feet. Various tests may also be done to help diagnose Marfan syndrome. These tests look for common complications of the disorder.

An echocardiogram, or echo, checks the heart to see how well its chambers and valves are working. This test uses sound waves to create pictures of the heart and its arteries. Imaging tests such as an MRI or CT scan may also be used. These tests can check the heart's arteries and the spine for signs of Marfan syndrome.

An MRI uses strong magnets to create images of the inside of the body. A CT scan is an x-ray machine linked to a computer. A CT scan takes detailed pictures of your organs.

An ophthalmologist, or eye specialist, may perform some eye exams to help diagnose Marfan syndrome. One of these is called a slit-lamp exam. This test uses a microscope with a light to check the eyes for problems.

Genetic testing can help diagnose Marfan syndrome. In general, genetic testing involves blood tests to detect changes in genes. However, many different genetic changes can cause Marfan syndrome. Short of mapping a patient's genes, there is no single blood test that can diagnose the disorder.

Doctors use a set of guidelines to help diagnose the disorder. Your symptoms, your family medical history, and the results of medical tests factor into the criteria.

Last reviewed: 12/18/2018

If you are diagnosed with Marfan syndrome, your healthcare provider may recommend your immediate family be checked as well. This is because the symptoms of Marfan syndrome can vary and another person in your family may have Marfan syndrome and not know it. Genetic testing and counseling are often very helpful in the work up of this disorder.

Treatment

There is no cure for Marfan syndrome. However, treatments can help delay or prevent complications linked to Marfan syndrome. Treatment depends on a person's symptoms.

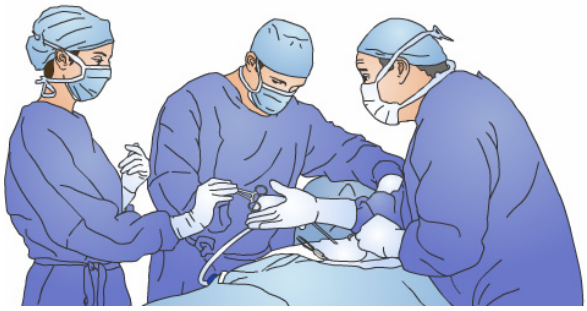
If you have Marfan syndrome, routine care and tests are needed to check the heart valves and aorta. If a problem is found, treatment may include medicines called beta blockers to relieve strain on the aorta.

Surgery may also be needed to repair or replace a part of the aorta if it stretches or tears. After surgery, medicines called blood thinners can be prescribed to prevent blood clots from forming. These may be taken temporarily or for the rest of your life.

People with Marfan syndrome may also need bone and joint treatments. For example, a curve in the spine called scoliosis can be treated with a brace or surgery. Surgery may also be needed to repair a chest that sinks in or sticks out. This prevents the chest from pressing on the lungs and heart.

Glasses or contacts may help with some eyes problems caused by Marfan syndrome. Other eye problems could require surgery.

A problem with the brain and spinal cord called dural ectasia is common in people with Marfan syndrome. It is usually treated with pain medications.



Sometimes Marfan syndrome can also cause lung problems, such as a collapsed lung. To treat this condition, a tube is placed through the skin and chest wall to remove air between the chest wall and the lung and re-inflate the lung. Surgery may be needed.

Last reviewed: 12/18/2018

Summary

Marfan syndrome is a disorder that affects connective tissue. Most people with Marfan syndrome have heart and blood vessel problems. They may also have problems with their bones, eyes, skin, nervous system, and lungs.

Marfan syndrome is often inherited. This means that the defective gene that causes Marfan syndrome is passed on from parent to child. If one parent has the defective gene, he or she has a 50-50 chance of giving it to their child.

Marfan syndrome can be difficult to diagnose. Many different tests are needed. Doctors use a set of criteria and the results of medical tests to diagnose Marfan syndrome.

There is no cure for Marfan syndrome. Treatment focuses on delaying or preventing complications linked to Marfan syndrome.

Last reviewed: 12/18/2018