How does Marfan syndrome affect the aorta? and What are the Treatments?

What is marfan syndrome?

Marfan syndrome is a relatively common genetic disorder of connective tissue that affects many organ systems, but the most serious complications are aortic aneurysms and dissection. A variety of medical and surgical approaches are available to manage cardiovascular complications. Our aim was to compare elective graft surgery, elective valve-sparing surgery, and medical management of patients with Marfan syndrome and thoracic aorta disease on the basis of life expectancy with different aortic root diameters and rate of increase in aortic root size.
People with Marfan syndrome are usually tall and slender with disproportionately long arms, legs, fingers, and toes. The damage from Marfan syndrome can be mild or severe. If the aorta - the large blood vessel that carries blood from your heart to the rest of your body - is affected, the condition may become life-threatening.
Treatment usually includes medications to keep blood pressure low to reduce pressure on the aorta. Regular monitoring to check damage progression is vital. Many people with Marfan syndrome eventually need preventive surgery to repair the aorta.

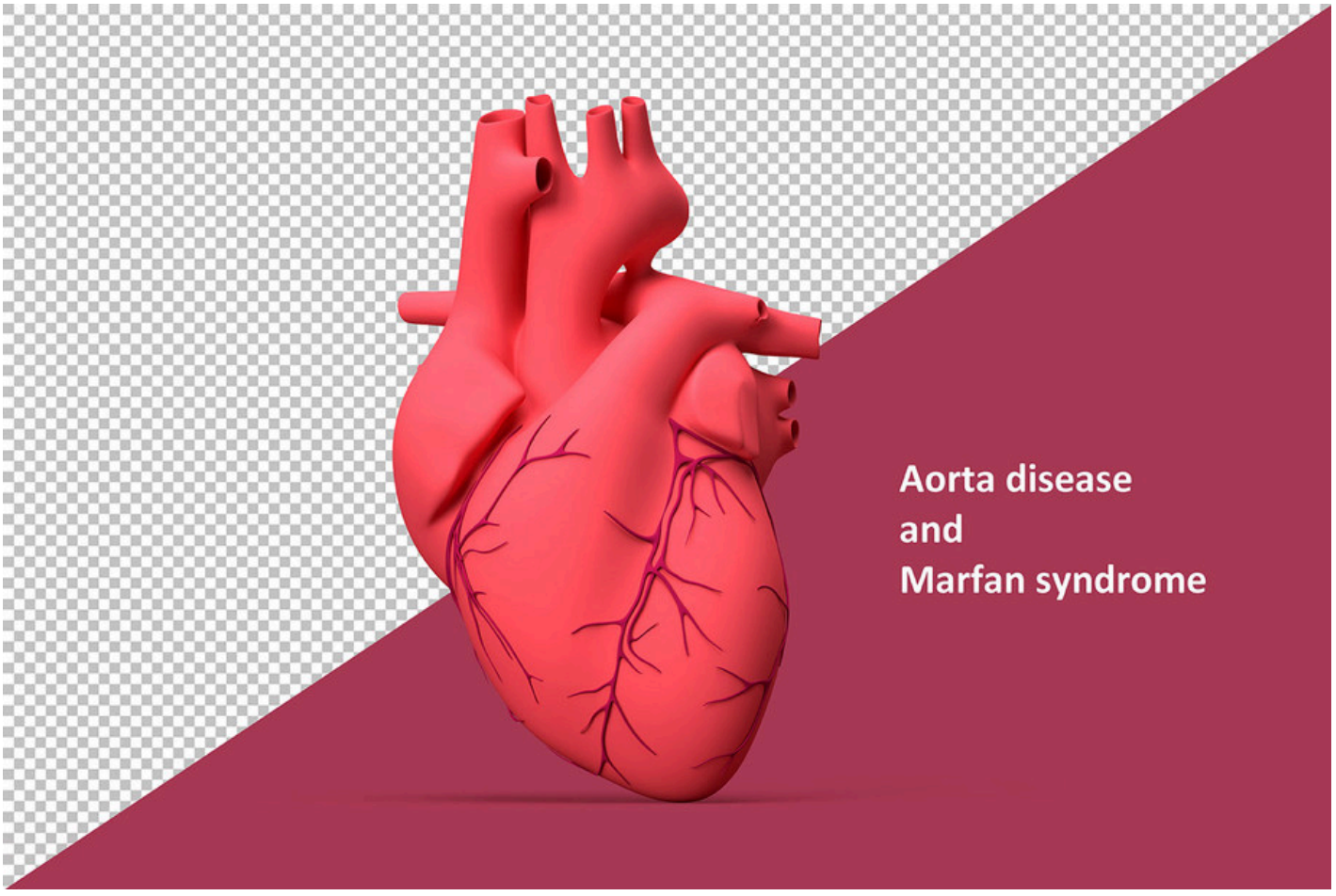
Marfan syndrome symptoms

Marfan syndrome is an inherited "altered expression" disorder. This means that signs and symptoms can vary from person to person. They can also vary in their severity, and they can range from mild to life-threatening. Symptoms tend to get worse with age.
People with Marfan syndrome may have:

- A tall, thin build
- A chest that sinks in or sticks out
- Crowded teeth
- Flat feet
- Disproportionately long arms, legs, fingers, toes, along with flexible joints
- The curvature of the spine(scoliosis)
- Heart murmurs
- Stretch marks

What causes marfan syndrome?

Marfan syndrome is rare, occurring in about 1 in 5,000 people. The Marfan disorder is brought about by a transformation in a quality called FBN. As far as possible the body's capacity to make the proteins expected to construct connective tissue. One out of four individuals with Marfan disorder will build up the condition for obscure reasons.



How is marfan syndrome diagnosed?

Marfan syndrome signs and symptoms vary from patient to patient. Here is a portion of the tests your primary care physician may perform:

- A physical examination and family history.
- With a stethoscope, the doctor will listen to your heart for any abnormal heart sounds.
- Echocardiography can be used to find valve function, movement of the heart wall and total heart volume.
- A computed tomography (CT or CAT) scan reveals cross-sectional images of the body, particularly the aorta.
- Magnetic resonance imaging (MRI) gives doctors a detailed image of the heart and aorta.

Treatment of marfan syndrome

There is no medical treatment to reverse the fibrillin abnormalities in people with Marfan syndrome. In the future, research on a strain of mice that bred with similar problems with fibrillin may lead to the successful treatment of Marfan.
Until then, doctors try to prevent or delay the changes in the aorta that appear in Marfan patients by prescribing drugs that reduce the pressure inside the aorta. Your doctor may advise you to take a beta-blocker and/or an angiotensin receptor blocker called losartan (Cozaar). Beta-blockers, such as propranolol (Inderal), metoprolol (Lopressor), atenolol (Tenormin) and reduce pressure on the aorta wall by slowing the heart rate and reducing the strength of heart contractions, especially during exercise. Losartan and other angiotensin receptor blockers help lower blood pressure, which reduces pressure on artery walls.
If you have Marfan, consult with a cardiologist doctor immediatly and your doctor will monitor your heart health with periodic echocardiograms to check for problems with the aorta and mitral valve. In the event that a major issue is discovered, you may require a medical procedure to supplant the aortic valve, mitral valve, or part of the aorta. Doctors are concerned when the size of the aortic root measured on an echocardiogram enlarges over time. Once it reaches 5 cm or more, many experts recommend surgery. Some recommend smaller aortic root enlargement surgery.
You should follow a non-strenuous exercise program that includes non-competitive, non-contact sports (walking, cycling, and jogging), which you can do at your own pace.
If you have Marfan-related scoliosis with a temperature between 20 and 40 degrees, you can be treated with a brace and physical therapy. For scoliosis greater than 45 degrees, you will need surgery. You should have an annual eye exam to look for Marfan-related eye problems. If you suffer from ectopia, you may be able to be treated with special lenses called lenses and special eye drops to dilate the pupil instead of surgery. If eye surgery is necessary, it should be performed in an ophthalmology center that specializes in treating Marfan syndrome.

Complications of marfan Syndrome

Marfan syndrome causes a variety of health complications. Many of these factors affect the valves of the heart and blood vessels, which is why we created our comprehensive care center.

Serious complications

The most serious complications may include the heart valves or the aorta, which is the main artery that supplies blood to your body. As a result, Marfan can lead to:

- **Aortic aneurysm:** When part of the aorta wall stretches and weakens, it is called an aortic aneurysm.

- **Aortic dissection:** Dilated and weakened aorta may rupture and leak blood. This serious condition, called an aortic dissection, causes intense pain in the front or back of the chest or abdomen.
- **Aortic regurgitation:** The aortic valve stretches and allows blood to leak back into the left ventricle, a type of aortic valve disease. Ultimately, this condition can cause cardiomyopathy (enlarged and weakened heart muscles) or congestive heart failure.
- **Mitral valve disease:** Marfan syndrome can cause two types of mitral valve disease. Mitral valve prolapse results when the valve between the upper and lower chambers on the left side of the heart does not close properly. As the condition progresses, it can cause mitral valve regurgitation, resulting in a heart murmur or heart palpitations, shortness of breath, and fatigue.

Other complications of marfan syndrome

Marfan syndrome can also cause these other complications.
These may include eye complications:

- Detached retina
- Early glaucoma or cataracts
- Dislocated lens
- Severe nearsightedness

Symptoms that affect other parts of the body may include:

- Dilation of the cyst around the spinal cord, which leads to pain, numbness, or weakness in the legs
- The tiny air sacs dilate in the lungs, which can lead to lung collapse

Risk factors

Marfan syndrome affects both men and women and occurs among all races and ethnic groups. Because it is an inherited condition, the biggest risk factor for Marfan syndrome is that a parent has the disorder.

Marfan syndrome prevention

For the prevention of complications, as well as improving the quality of life of patients diagnosed with MFS, careful monitoring and pharmacological and/or surgical intervention when indicated is critical.

- **Monitoring and precautions**

Cardiac abnormalities are common in patients with MFS and therefore, annual or more frequent echocardiograms are recommended to prevent fatal complications. Avoiding contact sports and activities that cause fatigue is extremely important for MFs because of the underlying weakness of the blood vessels and problems of the aorta. Additionally, it is important to use SBE (subacute bacterial endocarditis) with antibiotics during dental work in order to prevent bacterial infection of the heart valves or the structures inside the heart. Due to the fact that the symptoms of MF and other genetic disorders often overlap, genetic testing is valuable to confirm the diagnosis and facilitate the development of correct preventive measures and treatments.

- **Pharmacologic therapy**

Prescription medications such as beta-blockers or angiotensin receptor blockers that have the ability to reduce hemodynamic pressure on the aortic wall and slow the growth of the aortic root can improve the overall survival of those diagnosed. It also lowers blood pressure, reduces susceptibility to distention at the level of the ascending aorta, and reduces the rate of aortic complications. This treatment has been shown to be most beneficial when the diagnosis and appropriate treatment are started early.