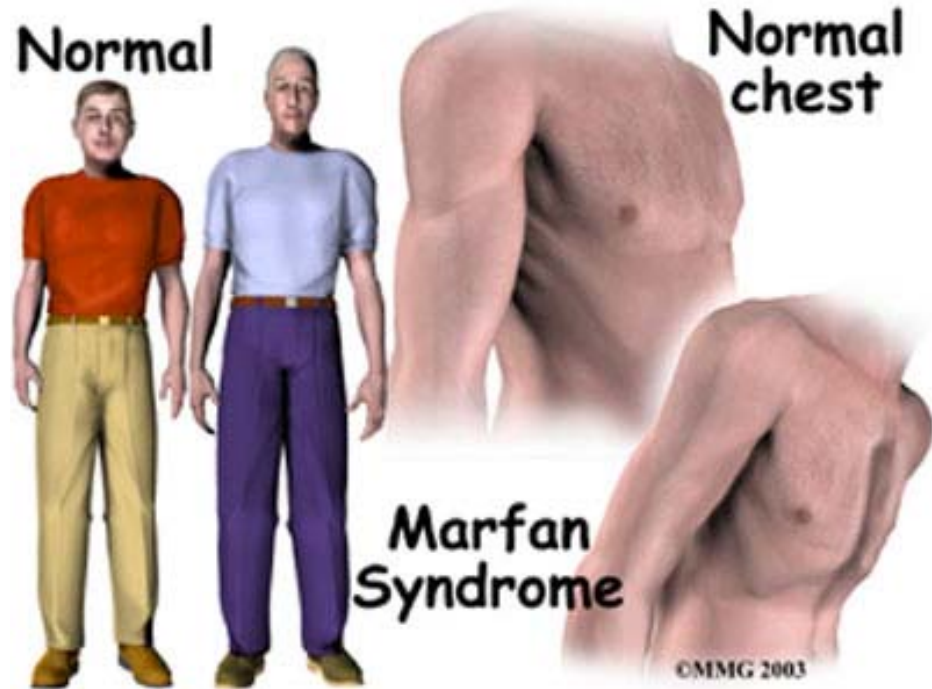


What is Marfan syndrome?

Marfan syndrome is heritable disorder of connective tissue caused by a mutation of a gene on the 15th chromosome. It is autosomal dominant so there is a 50% chance of parents passing it on to his/her offspring [1]. The connective tissue is defective and leads to abnormalities of a patient's physical appearance as well as the skeleton, eyes, heart and blood vessels, nervous system, skin and lungs.

What are the symptoms/characteristics?

- Disproportionally long limbs
- Long, thin fingers
- Typically tall stature
- Predisposition to cardiovascular abnormalities
- Abnormal curvature of the spine
- Nearsightedness
- Abnormal indentation of the sternum [3]



How many people have Marfan syndrome?

In the United States, approximately 1 in 5000 people have this condition. [3]

Marfan syndrome has no bias with regard to sex, ethnicity, or geography. It can affect anyone.

How can it be diagnosed?

There is no distinct test that can diagnose Marfan's. A physical exam by a doctor is the only way to determine if one has this syndrome. The doctor's findings usually lead to examination by specialists in bones, eyes, and the heart. Also, geneticists are consulted to determine if this hereditary difference has been seen in other family members.

If Marfan syndrome can be found in the family, two of the unique characteristics previously described are enough to diagnose Marfan's. If there are no signs of it in the family, three characteristics are needed to complete the diagnosis.

What happens during the testing process?

Family information is given to see if anyone else has this disorder or if there was an early, unexplained, heart related death. During the physical part of the examination, the body is measured to see the ratio between arm/leg to trunk. Then an ophthalmologist, who specializes in the eye, performs an eye examination. The weakening of the connective tissue can lead to the detachment of the retina. A cardiologist would administer heart tests such as an echocardiogram.

Are there any treatments?

There is no cure for Marfan syndrome but there are treatments available that can minimize, and in the best case, eliminate complications.

- Skeletal- Annual skeletal exams are to be given to see if there is an increased change in the spine or sternum. If so, a brace or even surgery can correct the problem. If not fixed, deformity can arise along with malfunction of the heart and lungs.
- Eyes – Along with the skeletal exams, eye exams must be given regularly. Corrective lenses can help but in serious cases, surgical options must be considered. The picture below shows a 34 year old man and his 10 year old son presenting nasal superior lens subluxation. This can cause retinal detachment and is associated with glaucoma.[1]



- Heart – Marfan syndrome predisposes to a rupturing or tearing of the aorta or other heart valves. Regular checkups and echocardiograms can make sure one's heart and aorta are safe from this. If a test reveals strain on the aorta, surgery can be performed to repair it.

Is there anything I can do to reduce the risks?

A patient with Marfan syndrome must not smoke. The increased heart and lung problems due to smoking can further damage the organs. Also, high intensity sports and physical activity which greatly elevates the blood pressure should be avoided. Increased heart rate can also increase the risk of rupturing a heart valve. Drugs can be given to reduce stress on the aorta.

What is the prognosis for the future?

While there is little hope for a total cure, much has been done to make people afflicted with this disorder live a normal life. Increases in awareness and detection allow for one to get help early on, before any serious problems arise. The average life span of someone with Marfan syndrome is about equal to a normal person.

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