

STANDING UP TO MARFAN SYNDROME

Features of Marfan Syndrome are Most Often Found in the Heart, Blood Vessels, Bones, Joints and Eyes

There is a 50% Chance That a Person with Marfan Syndrome Will Pass Along the Genetic Mutation to their Children

Marfan Syndrome is a genetic condition that affects the connective tissue in various parts of the body. Connective tissue holds all the body's cells, organs and tissue together. It also plays an important role in the body's growth and development.

How does Marfan Syndrome affect the body? What are the symptoms? And how is it treated? Please read the following summary to learn the answers to these questions.

01 | Understanding Marfan Syndrome

Connective tissue is made up of proteins. Due to a genetic defect, Marfan Syndrome patients have an increase in a protein called transforming growth factor beta. This protein imbalance leads to the features and symptoms of Marfan Syndrome, which include:

- Long arms, legs and fingers and flat feet
- Tall and thin body type
- Curved spine
- Sunken or protruding chest
- Hyper-flexible joints
- Crowded teeth
- Extreme nearsightedness
- Heart murmurs

02 | Complications

Because Marfan Syndrome can affect almost any part of the body, a variety of complications can arise, some of which are serious. The most dangerous complications involve the heart and blood vessels.

Examples include:

- **Aortic aneurysm**- The wall of the aorta weakens and bulges, and can sometimes rupture.

- **Aortic dissection** - This occurs when a small tear of the innermost layer of the aorta's wall allows blood to squeeze between the inner and outer layers of the aorta.
- **Valve malformations** - Heart valves can be overly elastic or malformed. This causes the heart to work harder and can result in heart failure.

03 | Diagnosis and Treatment

Orthopedic Specialists perform a thorough physical examination and medical history. Patients can also expect a number of tests to examine different areas of concern, such as heart tests to check the condition of the heart valves and aorta and eye tests to look for lens dislocation, cataracts, glaucoma or detached retina.

Treatment generally includes hypertension medication to prevent aorta enlargement and eyeglasses or contact lenses for vision problems. Surgical procedures may be required in severe cases.

For more information on Marfan Syndrome, please visit: <https://www.marfan.org/>

Did You Know?

About 1 in 5,000 people have Marfan Syndrome

References

<https://www.marfan.org/about/marfan>

<http://www.mayoclinic.org/diseases-conditions/marfan-syndrome/symptoms-causes/syc-20350782>

<https://ghr.nlm.nih.gov/condition/marfan-syndrome>