Aortic Disease and Marfan

What is the aorta?

The main artery that carries blood away from the heart. The blood leaves the heart through the aortic valve. Then it travels through the aorta, making a cane-shaped curve that allows other major arteries to deliver oxygen-rich blood to the brain, muscles and other cells.

Marfan syndrome

Marfan syndrome is a rare genetic disorder that makes the body's connective tissues—the ligaments, tendons, blood vessels, heart valves, cartilage, and more that bind various elements of the body together—weaker than they should be. This condition affects the development and functioning of the heart, blood vessels, eyes, skeleton, and other areas.

How common is Marfan syndrome, and how would I get it?

Marfan syndrome is very rare, with only 50,000 people diagnosed in the United States per year. Because it is caused by a genetic disorder, it is thought to be an inherited disease that can be passed down.

What impact does Marfan syndrome have on the heart and blood vessels?

The genetic defect that causes Marfan syndrome decreases the production of a crucial protein, fibrillin. Having insufficient fibrillin weakens the major arteries and makes the heart more susceptible to several serious conditions, including:

- **Aortic aneurysm** – a bulging of the weakened aorta, which can lead to rupture
- **Aortic dissection** – a tearing of the aorta that allows blood to leak through the tears and in between the layers of the aortic walls
- **Regurgitation** – a backward leak of blood caused by a weakened aortic valve, which can reduce the amount of blood reaching your organs and cause your heart to work much harder
- **Mitral valve prolapse** – a bulging of the mitral valve as the heart is pumping, which can lead to chest pain and heart palpitations
- **Increased chance of heart attack**
- **Irregular heart beat** (arrhythmia)
- **Sudden cardiac death**

How does Marfan syndrome present itself?

The symptoms described above will reveal themselves over time, with only 40% to 60% of patients having mitral valve prolapse or aortic issues. More obvious signs of Marfan syndrome include:

- A tall, thin frame with slender, long fingers, arms, and legs
- A curved spine (scoliosis)
- A concave or protruding breastbone
- Vision problems, including nearsightedness or a detached retina
- Flat feet
- Loose joints or being double jointed
- Shortened muscles, tendons, and ligaments (contracture)

How can I find out if I have Marfan syndrome?

There are many ways to detect this disorder. Your doctor will probably start with a family history and a physical examination to look for irregular heart sounds. If necessary, you may undergo further in-depth testing: an echocardiogram shows your valve function, heart wall motion, and overall heart size; a computed tomography (CT or "CAT") scan produces crosssectional images of your body, particularly the aorta; and magnetic resonance imaging (MRI) provides a more detailed look at your heart and aorta.

What is the treatment for Marfan syndrome?

For starters, regular doctor's visits will become a routine part of your life. These visits will include chest X-rays and echocardiograms to monitor your heart and how it is working. Depending on how severe your case is, your treatment may be as simple as taking beta-blockers to lower your heart rate and blood pressure, or it may require more extreme measures, such as surgery to mend your aorta or your heart valves.

How will Marfan syndrome affect my day-to-day life?

The prognosis for most individuals with Marfan syndrome is good, with many living late into their 60s with little more than a few extra doctor visits. That said, the following restrictions generally apply:

- Always consult with your doctor before you undergo any dental procedures or surgery. Precautionary steps may be needed to prevent bacteria from entering the bloodstream and causing an infection in your heart valves—for example, taking an antibiotic before the procedure.
- Intense workout regimens or contact sports may be prohibited.
- Women must consult a doctor before getting pregnant.