

What is Marfan's Syndrome?

Marfan's Syndrome is a condition caused by abnormal connective tissue in the body that can be inherited. Connective tissue is the "cement" or foundation that holds the body together. Abnormal or weak connective tissue can cause many different problems in the body. It can affect the heart and blood vessels, the lungs, the eyes, the skin, and the skeleton. The most serious problem is related to the heart. When the wall of the heart or the aorta (big vessel coming out of the heart) is weak it can stretch and eventually tear. When it tears it can cause a sudden loss of a lot of blood and sudden death. The most common place for the aorta to tear is in the first few inches next to the heart. It can tear at any point along the Aorta. Another common problem is that the valves of the heart can stretch and leak. The most commonly affected valve is the Mitral valve. Sometimes it can leak so much that it needs to be replaced.

There is no clear-cut way to diagnose Marfan's Syndrome. We decide that someone has Marfan's based primarily on physical findings. Patients with Marfan's Syndrome tend to be tall and thin. The span of their arms is greater than their height. The fingers and toes are long. They have loose joints and are often described as "double Jointed". They commonly have flat feet. They may have scoliosis (curvature of the spine) or have a chest wall abnormality causing the chest to be sunken in or protruding out. This is called Pectus excavatum or Pectus Carinatum. They tend to have a high palate in the mouth. The heart exam may show a heart murmur or a noise called a click. They tend to be near sighted and may have problems with the Retina (which is the part of the eye that actually allows us to see). The lens may be dislocated. Sometimes the skin may have "stretch marks" on it. We use the echocardiogram to diagnose the heart problems. Patients with Marfan's Syndrome have normal intelligence and development. About half of the time Marfan's Syndrome occurs as a sporadic event, in other words no one else in the family has it. It can be inherited from either the mother or father and can be passed on to children. We hope that in the future tests that look directly at connective tissue under a microscope will be available commercially to allow us to diagnose Marfan's Syndrome with more certainty. Also research is now underway to identify the genes that cause Marfan's and hopefully we will be able to identify carriers of the gene some day.

Treatment for Marfan's Syndrome is somewhat limited. If the Aorta stretches too much or tears it can usually be surgically fixed if caught in time. Valves that stretch too much can be replaced. Orthopedic problems such as scoliosis and pectus deformities can be fixed. Many times problems of the eyes can be fixed. If there is evidence of stretching of the Aorta we give a medication called Inderat or Tenormin (Beta-Blocker) to try and slow down the process of stretching of the aorta. This will not "cure" Marfan's Syndrome but can prevent some of the major problems.

Most people with Marfan's Syndrome can lead an active and productive life. But at least one third will have a serious problem with either the Aorta or the valves of their heart requiring surgery during their lifetime. Many Marfan patients need to refrain from doing vigorous or extreme sports, especially those that cause the blood pressure to rise significantly. People with Marfan's Syndrome need to always be followed by a doctor. Women with Marfan's Syndrome can have babies as long as there is no evidence of significant stretching of the Aorta. As a whole, people with Marfan's Syndrome have a slightly shorter life expectancy than normal.

If you have any questions please ask one of the doctors.