

Marfan's syndrome

Marfan's syndrome is a congenital disease that affects a type of collagen that the body produces. Collagen is a component of connective tissue. Connective tissue makes up the supporting structure of many of the body's muscular structures and ligaments. People with Marfan's syndrome have very elastic connective tissue. The walls of the main arteries are weak because the muscle and connective tissue that make up the wall of the blood vessels are also weak and excessively elastic. The aorta, which is the main artery that comes out of the heart, tends to be affected.

Diagnosis of Marfan's syndrome

Marfan's syndrome is a hereditary trait that is characterized primarily by its musculoskeletal manifestations. Among these, tall stature, arachnodactyly (long fingers), excessively long arms in relation to height and articular hypermobility are frequent. The diagnosis must include at least three family history criteria, ocular symptoms that include serious myopia or luxation of the eye lens, and the musculoskeletal symptoms mentioned above.

Complications of Marfan's syndrome

As a result of the elasticity of the artery wall, the aorta can become enlarged, which can weaken the inner wall of the aorta even more. This increases the risk of rupture of the inner wall of the aorta. Blood can leak through these tears in the artery wall, separating its layers. This is called aortic dissection. Another problem that could occur is the weakening of the artery wall, which increases the likelihood of having an aneurysm (large protuberance on the wall of the aorta). Any of these problems increases the risk of rupture of the aorta. It is common for the aortic origin (the portion of the aorta that is joined to the heart) to be affected.

The abnormal collagen can also affect the function of any of the heart's four valves. The valves' greater elasticity could cause leakage. When this occurs, the heart has to work harder, since it has to pump blood in a forward and backward direction through the valve that is leaking. In the final analysis, this can cause the heart muscle to fail. When heart valves leak, they can cause an abnormal noise called heart murmur. Your doctor can hear it when he auscultates your heart.

Monitoring and treatment

Marfan's syndrome is a serious condition that requires periodic monitoring by a doctor familiar with the complications of this disease. Echocardiograms tend to be done to evaluate and monitor the heart. Other tests may be necessary to monitor the disease. Treatment is based on the state of the heart and circulatory system, as they appear in the diagnostic tests. Although some people do not need treatment, it is still necessary to examine them periodically. Some patients receive medications to lower blood pressure and the heart's workload in order to slow the progression of the disease. Others may need surgical repair or replacement of abnormal heart valves or weakened portions of the aorta.

The intensity of exercise recommended for patients with Marfan's syndrome is based on the state of their disease. People with dilation of the aorta should avoid any strenuous activity. Isometric exercises, such as weightlifting and rowing, and contact sports in which blows to the chest could be received should also be avoided in all people with diagnosed Marfan's syndrome.

Pregnancy is not advised in women with Marfan's syndrome who have dilation of the aorta, due to the risk of rupture of the aorta during pregnancy or delivery. Marfan's syndrome is an autosomal dominant disease, which means that a significant risk exists of transmitting the gene for the disease from parent to child. Consulting a genetic counselor is beneficial for people with Marfan's syndrome.