FBN1

The *FBN1* gene is one of many genes that helps provide strength and stability to tissues in the body. The *FBN1* gene makes a protein which is used to form elastic fibers that allow the skin, ligaments, and blood vessels to stretch. When this protein doesn't work properly, the tissues can be weakened, especially the blood vessels surrounding the heart.

Impact of *FBN1* mutations
Individuals with a mutation in the *FBN1* gene are at an increased risk for developing developing different hereditary cardiovascular (heart and blood vessel) disorders called arteriopathies, which can cause weakness, enlargement, and tears of the walls of the arteries.
These include familial thoracic aortic aneurysm and dissection and Marfan syndrome.

Disorders associated with the *FBN1* gene

Mutations in the *FBN1* gene have been associated with the following disorders:

Familial Thoracic Aortic Aneurysm and Dissection
Familial thoracic aortic aneurysm and dissection (FTAAD) is a hereditary disorder associated with problems with the large blood vessel that carries blood away from the heart to the rest of the body (aorta).

Individuals with FTAAD commonly have problems with the upper part of the aorta (thoracic aorta), which is located in the chest near the heart. The walls of the aorta can become weakened and stretch (aortic dilation). This can lead to a bulge in the wall of the aorta (aortic aneurysm) or a sudden tearing of the aorta (aortic dissection). Aortic aneurysm and aortic dissection can be life threatening.

In individuals with FTAAD, the age of onset and severity of symptoms may vary, even within the same family. Some individuals with FTAAD experience no noticeable symptoms, but are still at risk for aortic dissection. Women with FTAAD are at increased risk for serious and possibly life-threatening complications during pregnancy and may require special care.

Diagnosing FTAAD typically involves evaluating an individual's medical and family histories, as well as a regular physical exam, and an imaging test used to see the heart and aorta (echocardiogram). These evaluations may be combined with genetic testing to diagnose FTAAD.

Treatment for FTAAD typically includes frequent monitoring of the aorta to look for dilation and aneurysms. If an aneurysm is present, it may require surgical repair to prevent dissection. Certain medications can also be used to control blood pressure and reduce stress on the walls

of the aorta. It is generally recommended that individuals avoid strenuous exercise, contact sports, smoking, and a diet high in cholesterol.

Marfan Syndrome
Marfan syndrome is a hereditary disorder associated with problems with the structure of connective tissue in many parts of the body, which can cause weakness of the blood vessel walls, dislocation of the lens in the eye (ectopia lentis), and increased flexibility in the joints.

Marfan syndrome is a connective tissue disorder. Connective tissue supports, binds, or connects other tissues or organs in the body. Individuals with Marfan syndrome commonly have problems with the heart and the surrounding blood vessels, especially the large blood vessel that carries blood away from the heart to the rest of the body (aorta). The walls of the aorta can become weakened and stretch (aortic dilation). This can lead to a bulge in the wall of the aorta (aortic aneurysm) or a sudden tearing of the aorta (aortic dissection). Aortic aneurysm and aortic dissection can be life threatening. Other heart problems may include leaks in the valves that connect the chambers of the heart, which can cause fatigue, shortness of breath, and strong or irregular heartbeats (heart palpitations).

Individuals may have problems with the eyes, such as dislocation of the lenses in one or both eyes (ectopia lentis), nearsightedness, cataracts, or glaucoma. They may have a tall and slender body type with long fingers and toes (arachnodactyly). Other symptoms may include increased flexibility in joints, a curved spine (scoliosis), back pain, flat feet, collapsed lung (pneumothorax), and a sunken chest (pectus excavatum) or a protruding chest (pectus carinatum). The features of Marfan syndrome may appear at any age. Women with Marfan syndrome are at increased risk for serious and possibly life-threatening complications during pregnancy and may require special care.

Diagnosing Marfan syndrome typically involves evaluating an individual's medical and family histories, as well as a regular physical exam, and an imaging test used to see the heart and aorta (echocardiogram). These evaluations may be combined with genetic testing to diagnose Marfan syndrome.

Treatment for individuals with Marfan syndrome may include medications that help control blood pressure to reduce stress on the walls of the aorta, as well as frequent echocardiograms to identify aneurysms that develop. If aneurysms develop, surgery may be required. Some individuals may also need surgeries to correct a curved spine or a dislocated lens in the eye. It is also recommended that individuals avoid contact sports or strenuous exercise and substances that stimulate the heart, such as certain cold medicines and caffeine.

Useful resources

American Heart Association
Focused on building lives free of heart disease by providing accessible education and funding innovative research.
http://www.heart.org

The John Ritter Foundation for Aortic Health
Dedicated to improving the identification of individuals at risk for aortic dissections and the treatment of thoracic aortic disease through medical research.
http://johnritterfoundation.org

The Marfan Foundation
Provides information and support to healthcare providers, caregivers, and families affected by Marfan syndrome and related disorders, including FTAAD.
http://www.marfan.org

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