COL3A1

The *COL3A1* gene is one of many genes that helps provide strength and stability to tissues in the body. The *COL3A1* gene makes a protein which is found in the skin, lungs, and the blood vessels. When this protein doesn't work properly, these tissues can be weakened, especially the blood vessels surrounding the heart.

Impact of *COL3A1* mutations
Individuals with a mutation in the *COL3A1* gene are at an increased risk for 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 vascular Ehlers Danlos syndrome.

Disorders associated with the *COL3A1* gene

Mutations in the *COL3A1* 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.

Vascular Ehlers-Danlos syndrome
Vascular Ehlers-Danlos syndrome (vEDS) 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 and other organs, fragile skin, and easy bruising and bleeding.

vEDS is a connective tissue disorder. Connective tissue supports, binds, or connects other tissues or organs in the body. Individuals with vEDS commonly have problems with the heart and the surrounding blood vessels, especially the large blood vessels that carry blood away from the heart to the rest of the body (arteries). The walls of the arteries can become weakened and stretch (dilation). This can lead to a bulge in the wall of the artery (aneurysm) or a sudden tearing of the artery (dissection). In individuals with vEDS, dissections commonly occur in the aorta, the largest artery in the body, but may occur in other arteries. Arterial dissections are life threatening, and are the major cause of death in individuals with vEDS .

Other common features of vEDS include easy bruising and bleeding, enlarged veins, and very flexible joints (joint hypermobility). Some individuals may have specific facial features, as well as thin, clear skin that wrinkles on the hands and feet. Besides dissection of the arteries, vEDS is associated with other major complications, including ruptures in the gastrointestinal tract, spleen, or liver, as well as collapsed lung (pneumothorax). This can lead to internal bleeding, stroke, and shock. Women with vEDS are at increased risk for serious and possibly life-threatening complications during pregnancy, such as rupture of the uterus, and may require special care.

Diagnosing vEDS 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 vEDS.

Treatment for vEDS is typically focused on preventing complications. Individuals are recommended to have a heart exam that includes an echocardiogram. They may also benefit from wearing protective pads or bandages to prevent bruising and bleeding. It is usually recommended that individuals with vEDS avoid contact sports and strenuous exercise, as well as certain medications that keep the blood from clotting. Surgery and other invasive procedures are generally discouraged, except if necessary and with special precaution.[1]

Useful resources

EDS Network Cares
Improves the quality of life for people who have Ehlers-Danlos Syndrome through research, education, and support.
http://www.ehlersdanlosnetwork.org

The Ehlers-Danlos Society
Guides both patients and medical professionals to information, resources, support, and education.
https://ehlers-danlos.com

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

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[1] Malfait F, De paepe A. The Ehlers-Danlos syndrome. Adv Exp Med Biol. 2014;802:129-43.