

## ACTA2

The *ACTA2* gene is one of many genes that helps provide strength and stability to tissues in the body. The *ACTA2* gene makes a protein which prevents arteries from stretching out as blood is pumped through them. When this protein doesn't work properly, the tissues can be weakened, especially the blood vessels surrounding the heart.

### Impact of *ACTA2* mutations

Individuals with a mutation in the *ACTA2* gene are at an increased risk for developing familial thoracic aortic aneurysm and dissection (FTAAD), a hereditary cardiovascular (heart and blood vessel) called an arteriopathy. This disorder can cause weakness, enlargement, and tears of the walls of the arteries.

### Disorders associated with the *ACTA2* gene

Mutations in the *ACTA2* gene have been associated with familial thoracic aortic aneurysm and dissection (FTAAD).

#### **Familial thoracic aortic aneurysm and dissection (FTAAD)**

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.

## 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>

### **TAD Coalition**

Committed to increasing public awareness of the factors that put people at risk for aortic aneurysm and dissection, and to improving the diagnosis and management of these life-threatening conditions.

<http://www.tadcoalition.org>