Loeys-Dietz syndrome is caused by a genetic mutation of either one of the four genes that tell the body how to make proteins, including the proteins in connective tissue. These genes are TGFBR1, TGFBR2, SMAD3, TGFB2 and TGFB3. When any of these genes has a mutation, growth and development of the body's connective tissue and other body systems is disrupted, which can lead to the signs and symptoms of Loeys-Dietz syndrome.

One of the key features of Loeys-Dietz syndrome is an enlargement of the aorta, the large blood vessel that carries blood from the heart to the rest of the body. The aorta can weaken and stretch, causing a bulge in the blood vessel wall (an aneurysm). Stretching of the aorta may also lead to a sudden tearing of the layers in the aorta wall (aortic dissection). This is a life-threatening complication that can occur without warning. In Loeys-Dietz syndrome, aneurysms and dissections also can occur in arteries other than the aorta.

Some additional features of Loeys-Dietz syndrome include –

- Arteries that twist and wind – arterial tortuosity
- Heart birth defects – PDA, atrial septal defect, bicuspid aortic valve
- Wide spaced eyes
- Cleft palate
- Bifid uvula
- Instability of the spine in the neck
- Easy bruising, wide scars and 'velvet' translucent skin
- Allergies
- Gastrointestinal problems
- Rupture of hollow organs
- Some similar physical signs to Marfan syndrome

The above are a list of possible signs of Loeys-Dietz syndrome, but it should be noted that not all patients have all of them, nor do any lead concretely to a diagnosis of LDS. You should consult with your doctor if you have individual health concerns.

More information can be found at www.marfan.ie or www.loeysdietz.org

Marfan syndrome is a genetic disorder, caused by a mutation in FBN1 gene, that affects the body's connective tissue. Connective tissue holds all the body's cells, organs and tissue together. It also plays an important role in helping the body grow and develop properly.

Because connective tissue is found throughout the body, Marfan syndrome can affect many different parts of the body, as well. Features of the disorder are most often found in the heart, blood vessels, bones, joints, and eyes. Some Marfan features – for example, aortic enlargement (expansion of the main blood vessel that carries blood away from the heart to the rest of the body) – can be life-threatening. The lungs, skin and nervous system may also be affected.

Some additional features of Marfan syndrome include –

- Dislocated lenses, early glaucoma, early cataracts, detached retina, in the eyes
- Lung collapse – pneumothorax
- Long arms, legs and fingers
- Tall thin body type
- Curved spine
- Chest wall that sticks out or sinks in
- Flexible joints
- Flat feet
- Teeth overcrowding
- Stretch marks that are not related to weight gain or loss

Not everyone with Marfan syndrome has all of the above features. If you suspect Marfan you should consult with your doctor.

For more information see www.marfan.ie or www.marfan.org