LOEYS-DIETZ SYNDROME

Loeys-Dietz syndrome is a genetic disorder of the body's connective tissue. It has some features in common with Marfan syndrome, but it also has some important differences.

People with Loeys-Dietz syndrome features need to see a doctor who knows about the condition to decide if they have the disorder; often this will be a medical geneticist. It is very important that people with Loeys-Dietz syndrome get an early and correct diagnosis so they can get the right treatment.

The Loeys-Dietz Syndrome Foundation, a division of The Marfan Foundation, provides additional information for those affected by Loeys-Dietz syndrome.

What causes Loeys-Dietz syndrome?

Loeys-Dietz syndrome (type 1-5) is caused by a genetic mutation in one of five genes that encode for the receptors and other molecules in the transforming growth factor-beta (TGF-β) pathway. These genes are:

- LDS-1- transforming growth factor beta-receptor 1 (TGFβR1)
- LDS-2 - transforming growth factor beta-receptor 2 (TGFβR2)
- LDS-3 - mothers against decapentaplegic homolog (SMAD-3)
- LDS-4 - transforming growth factor beta-2 ligand (TGFβ2)
- LDS-5- transforming growth factor beta-3 ligand (TGFβ3)

When any of these genes has a mutation, growth and development of the body's connective tissue and other body systems is disrupted, leading to the signs and symptoms of Loeys-Dietz syndrome. Marfan syndrome is different from Loeys-Dietz syndrome in that the gene mutation which causes Marfan syndrome is in fibrillin-1 (FBN-1), a protein within the connective tissue in the body. However, there are many common features between the two syndromes.

To connect with others who experience Loeys-Dietz syndrome, and to access additional information, visit The Loeys-Dietz Syndrome Foundation, a division of The Marfan Foundation.

What are the key features of Loeys-Dietz syndrome?

Because connective tissue is found throughout the body, Loeys-Dietz syndrome features can occur throughout the body, too, including the heart, blood vessels, bones, joints, skin, and internal organs such as the intestines, spleen, and uterus.

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.

Other features of Loeys-Dietz syndrome include:

Cardiovascular

- Arteries that twist and wind (arterial tortuosity)
- Heart defects at birth (atrial septal defect, patent ductus arteriosis, bicuspid aortic valve)

Eyes, Head and Neck

- Widely-spaced eyes (hypertelorism)
- White of the eye looks blue or gray
- Wide or split uvula (the tissue that hangs down in the back of the throat)
- Cleft palate
- Instability or malformation of the spine in the neck

Skin

- Easy bruising, wide scars, soft skin texture, and translucent skin (when it looks almost see-through)

Bones

- Club foot (when the foot is turned inward and upward at birth)
- Poor mineralization of the bones (osteoporosis) that can make the bones more likely to break)

Other

- Allergies to food and elements in the environment
- Stomach and intestine problems, such as difficulty absorbing food and chronic (comes and goes but never really goes away) diarrhea, abdominal pain, and/or gastrointestinal bleeding and inflammation
- Rupture of the spleen or bowel
- Rupture of the uterus during pregnancy

When a person has these particular features, it is important to be evaluated for Loeys-Dietz syndrome.

For more information, head to The Loeys-Dietz Syndrome Foundation, a division of The Marfan Foundation. You may also contact our Help Center with questions anytime at LoeysDietz.org/Ask (Please include blue buttons, in addition to the hyperlinked text for this section – one for LDSF and one for Help Center)

To learn more about Loeys-Dietz syndrome and its diagnosis and treatment, download:

LOEYS-DIETZ SYNDROME

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