

HEALING HEARTS.
INSPIRING HOPE.

Loeys·DietzTM
Syndrome Foundation 

Loeys-Dietz syndrome (LDS) is a genetic disorder that affects the connective tissue in the body.

Individuals with Loeys-Dietz syndrome exhibit a variety of medical features in the cardiovascular, musculoskeletal, skin and gastrointestinal systems. Information about the natural history and management of individuals with LDS continues to evolve.

What Are the Characteristics of Loeys-Dietz Syndrome?

- Aneurysms throughout the arteries in the body; the aortic root is the most commonplace of enlargement
- Tortuous (winding) arteries, most often in the neck
- Widely-spaced eyes
- Bifid (split) or broad uvula
- Craniosynostosis
- Joint laxity
- Hernias
- Skeletal features: Clubfoot, scoliosis, pectus deformities
- Cervical spine instability
- Congenital heart defects
- Food and environmental allergies



What Causes Loeys-Dietz Syndrome?

Loeys-Dietz syndrome is inherited in an autosomal dominant pattern, meaning that if someone has LDS, each of their offspring has a 50% chance of inheriting the gene mutation. Many individuals are the first in the family to have LDS. In this case, a random mutation occurred in the egg or sperm during conception. LDS affects all ethnic groups and both sexes.

LDS is caused by mutations (gene changes) in one of the following genes:

- | | | |
|----------|---------|---------|
| • TGFBR1 | • SMAD3 | • TGFB3 |
| • TGFBR2 | • TGFB2 | • SMAD2 |



How is Loeys-Dietz Syndrome Diagnosed?

If there is a suspicion of LDS, or a personal or family history of aneurysm, a genetics evaluation should be considered. Early and accurate diagnosis can help in getting early treatment and surveillance.

Vascular imaging (echocardiogram, MRA/CTA imaging) may also be performed.

Contact your primary care provider to obtain genetic testing for Loeys-Dietz syndrome and better comprehend the advantages and limitations of the testing process and the results.

How is Loeys-Dietz Syndrome Managed?

Your physicians will help shape your treatment and surveillance plan. This commonly includes:

- Serial vascular imaging to identify/assess aneurysm presence or growth
- Blood pressure medication and activity restrictions to reduce stress on aorta/arteries
- Surgical intervention when indicated

CONSULTATION AND TREATMENT BY VARIOUS SPECIALTIES:

- Cardiovascular surgery
- Allergy
- Gastroenterology
- Pulmonary
- Ophthalmology

Cardiac Emergencies: Know the Signs of an Aortic Dissection

Symptoms of an aortic aneurysm may be related to the location, size, and growth rate of the aneurysm and can include:

- Pain in the chest, neck, and/or back
- Swelling of the head, neck, and arms
- Coughing, wheezing, or shortness of breath
- Coughing up blood

Symptoms of aortic dissection usually appear suddenly and may include:

- Severe, sudden, constant chest pain and/or upper back pain, sometimes described as “ripping” or “tearing”
- Pain that feels like it is moving from one place to another
- Unusually pale skin
- Faint pulse
- Numbness or tingling
- Paralysis
- In some instances, there may be no pain but a sense that there is something terribly “wrong.”

If a dissection is suspected, a person needs immediate medical attention.

Download the LDS Emergency Preparedness Kit





Our Mission

- Encouraging education about Loeys-Dietz syndrome (LDS) and related connective tissue disorders to medical professionals and lay communities in order to aid in identification, diagnosis, and treatment of LDS
- Fostering research about LDS
- Providing a support network for individuals, parents, and families affected by LDS

Join the Loeys-Dietz Syndrome Community!

- Annual Conference
- Loeys-Dietz Awareness Month in June
- Empowerment Series Webinars
- Virtual Support Groups
- Walk for Victory

For more information, visit www.loeysdietz.org



The Loeys-Dietz Syndrome Foundation is a Division of The Marfan Foundation. *Learn more about The Marfan Foundation's fight for victory over genetic aortic and vascular conditions at marfan.org*