

Loeys-Dietz syndrome

A Correct Diagnosis

How To Find Out If You Have Loeys-Dietz syndrome

In 2005, doctors identified and named a newly defined genetic connective tissue disorder called Loeys-Dietz syndrome (LDS). For some people, a diagnosis of LDS can now explain why they have certain features and medical problems. Here is what you need to do to find out if LDS is the correct diagnosis for you or a family member.

Find a doctor who knows about Loeys-Dietz syndrome. Because Loeys-Dietz syndrome is newly identified, not all doctors are aware of it. You need to find a doctor who knows about LDS and how to tell the difference between LDS and the other connective tissue disorders.

The type of doctor most likely to know about LDS is a medical geneticist (a doctor who specializes in genetic disorders including Marfan syndrome (MFS), Ehlers-Danlos syndrome (EDS) and LDS).

You can find a medical geneticist by:

- asking your primary doctor for a referral
- calling the doctor referral service at your local hospital
- using online resources including American College of Medical Genetics (www.acmg.net) or National Society of Genetic Counselors (www.nsgc.org)

List The Features Of LDS Not Common In Other Connective Tissue Disorders

Write down any of these features you or your family members have. They include:

- Arterial tortuosity (twisting or spiraled arteries)
- Aneurysms and dissections in arteries other than the aorta
- Heart defects at birth such as atrial septal defect, patent ductus arteriosus, bicuspid aortic valve
- Hypertelorism (widely-spaced eyes)
- Blue sclerae (blue tinge to the whites of the eyes)
- Bifid (split) or broad uvula (the little piece of flesh that hangs down in the back of the mouth)
- Cleft palate (a gap in the roof of the mouth)
- Club foot (when the foot is turned inward and upward at birth)
- Gastrointestinal 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
- Allergies to food and things in the environment
- Cervical-spine instability (instability in the vertebrae directly below the skull)
- Osteoporosis (poor mineralization of the bones) that can make the bones more likely to break

List The Features Of LDS Found In Other Connective Tissue Disorders

Write down any of these features you or your family members have. They include:

- Aortic dilation or aneurysm (enlarged or bulging aorta, the main blood vessel that carries blood from the heart)
- Aortic dissection (tear of the wall of the aorta)
- Mitral Valve Prolapse – MVP (“floppy” mitral valve)

List The Features Of LDS Found In Other Connective Tissue Disorders Cont.

- Pectus excavatum (chest wall deformity that pushes the sternum and breast bone inward) or Pectus carinatum (chest wall deformity that pulls the sternum and breast bone out)
- Scoliosis (s-like curvature of the spine) or Kyphosis (spine that curves from back to front)
- Flexible joints
- Flat feet
- Features in the skin such as: easy bruising, wide scars, soft skin texture, and translucent skin (almost see-through)
- Rupture of the spleen or bowel
- Rupture of the uterus during pregnancy

Take Your Lists To Your Doctor

Explain the reasons why you think you might have LDS to your doctor. Also take lists of:

- your past illnesses, operations, and hospitalizations
- medications you are taking
- family members who have, or might have, LDS

Have your doctor do a thorough physical exam. This includes looking for LDS features in your bones, joints, skin, and face.

Talk To Your Doctor About Ordering Medical Tests.

These tests include:

- Echocardiogram. This ultrasound looks at the heart, its valves, and the aorta (vessel that carries blood from the heart) close to the heart.
- Either a computerized tomography (CT) or magnetic resonance (MR) angiogram (study of the blood vessels) with 3-D reconstruction, from the top of the head to the pelvis. The CT or MR imaging can find twisted blood vessels and aneurysms in other blood vessels in the body. These features are common in LDS and help doctors both make the diagnosis and plan the right medical care.
- Genetic testing that can find the TGFBR1, TGFBR2, SMAD3 or TGFB2 mutation (genetic change) that causes LDS. This testing is most helpful when used in people who have LDS features not usually seen in other connective tissue disorders. Your doctor must order this testing and they may refer you to a medical geneticist.

When genetic testing finds a gene mutation in one of these genes, the person most likely has LDS and needs special medical care and counseling. Ask your doctor how to take care of yourself. It is very important to follow these instructions. You also need to find out if there are other people in your family who also have LDS.

When genetic testing does not find a mutation it is still possible for a person to have LDS or a different connective tissue disorder. Ask your doctor if you have any other condition that needs medical care.

To Learn More About Loeys-Dietz syndrome

Read the other fact sheets on LDS. Find them online at the Loeys-Dietz Syndrome Foundation Web site at www.loeysdietz.org/resources.php.

