

How to Find Out if You Have Loeys-Dietz Syndrome



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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 (LDS)

Because Loeys-Dietz syndrome (LDS) is newly identified, not all doctors know about 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 kind 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
- calling the National Marfan Foundation Resource Center at 800-862-7326, ext. 26

GO OVER THE LIST OF LDS FEATURES THAT ARE NOT USUALLY SEEN IN OTHER CONNECTIVE TISSUE DISORDERS.

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

- Arteries that twist and wind (arterial tortuosity)
- Aneurysms and dissections in arteries other than the aorta
- Heart defects at birth such as atrial septal defect, patent ductus arteriosus, bicuspid aortic valve
- 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 (when the roof of the mouth is split at birth)
- Club foot (when the foot is turned inward and upward at birth)
- Gastrointestinal problems (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
- Allergies to food and things in the environment
- Malformation or instability of the spine in the neck (cervical spine instability)
- Poor mineralization of the bones (osteoporosis) that can make the bones more likely to break

GO OVER THE LIST OF FEATURES OF LDS THAT ARE ALSO FOUND IN OTHER CONNECTIVE TISSUE DISORDERS.

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

- Enlarged or bulging aorta, the main blood vessel that carries blood from the heart (aortic dilation or aneurysm)
- Tear of the wall of the aorta (aortic dissection)

Originally created in the U.S.A.
by



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- “Floppy” mitral valve (mitral valve prolapse – MVP)
- Chest that sinks in (pectus excavatum) or sticks out (pectus carinatum)
- Spine curves to the side (scoliosis) or from front to back (kyphosis)
- Flexible joints
- Flat feet
- Premature fusion of the bones in the skull (craniosynostosis)
- Swelling, bulging or widening of the spinal sac (dural ectasia)
- Features in the skin such as: easy bruising, wide scars, soft skin texture, and translucent skin (when it looks almost see-through)
- Rupture (tearing) of the spleen or bowel
- Rupture (tearing) of the uterus during pregnancy

TAKE YOUR LISTS TO YOUR DOCTOR.

Explain your reasons why you think you might have LDS. 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 test 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 top of the legs. The CT or MR 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 or TGFBR2 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. To find Laboratories that do LDS genetic testing go to the website of Gene Tests at www.genetests.org and select Laboratory Directory.

When genetic testing finds a either a TGFBR1 or TGFBR2 mutation, 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 a person has LDS or a different connective tissue disorder. Ask your doctor if you have any other condition that needs medical care. Also ask how you can get this care.

WAYS TO LEARN MORE

- Contact the Marfan Syndrome Support Group Ireland at info@marfan.ie.
- Talk to your doctor. Sometimes it helps to use information like this fact sheet when you speak with the doctor.
- Visit the Marfan Syndrome Support Group website at www.marfan.ie.
- Visit the Loeys-Dietz Foundation website at www.loeysdietz.org.