



Loeys-Dietz Syndrome Facts



The NMF gratefully acknowledges the Loeys-Dietz Syndrome Foundation for their partnership in creating this fact sheet. For more information about the Loeys-Dietz Syndrome Foundation, visit www.loeydietz.org.

WHAT IS LOEYS-DIETZ SYNDROME (LDS)?

Loeys-Dietz syndrome (LDS) is a genetic disorder of connective tissue that was identified and named in 2005. At that time doctors realized that even though LDS has some features of other connective tissue disorders, it is a distinct disorder. The other disorders that share features with LDS include Marfan syndrome, Ehlers-Danlos syndrome vascular type, and Shprintzen-Goldberg syndrome.

WHAT CAUSES LOEYS-DIETZ SYNDROME?

LDS is caused by a change (mutation) in either of the two genes that tell the body how to make proteins called transforming growth factor beta receptor 1 (TGFB1) and transforming growth factor beta receptor 2 (TGFB2.) When either gene is mutated, the transforming growth factor receptor does not function as it should and LDS features are the result.

WHO HAS LOEYS-DIETZ SYNDROME?

Because LDS is newly recognized, it is not known how many people have the disorder. It is likely there are people who are diagnosed with another connective tissue disorder who actually have LDS. This is especially likely for some people diagnosed with Marfan syndrome or “atypical” Marfan syndrome.

LDS affects both males and females. People can inherit LDS, meaning they get the mutation from a parent who has LDS. Others can have a spontaneous mutation, meaning they are the first in the family to have LDS.

WHAT ARE THE FEATURES OF LOEYS-DIETZ SYNDROME?

Because connective tissue is found throughout the body, LDS features can occur in the heart; blood vessels; bones; joints; skin; and internal organs such as the intestines, spleen, and uterus. Some LDS features are easy to see. Others features, such as heart and blood vessel problems, need special tests to find them.

Some LDS features are also found in Marfan syndrome . These 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)
- “Floppy” mitral valve (mitral valve prolapse – MVP)
- Chest that sinks in (pectus excavatum) or sticks out (pectus carinatum)
- Spine that curves to the side (scoliosis) or from the back to the front (kyphosis)
- Flexible joints
- Flat feet
- Swelling, bulging or widening of the spinal sac (dural ectasia)

Some LDS features are different from Marfan syndrome features and are very important for making a correct diagnosis. When a person has these particular features, it is important that the doctor think about LDS. Features that set LDS apart from Marfan syndrome and many other connective tissue disorders include:



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- Arteries that twist and wind (arterial tortuosity)
- Aneurysms and dissections in arteries other than the aorta
- Widely-spaced eyes (hypertelorism)
- Wide or split uvula (the tissue that hangs down in the back of the throat)
- Cleft palate
- Club foot (when the foot is turned inward and upward at birth)
- White of the eye looks blue or gray
- Heart defects at birth such as atrial septal defect, patent ductus arteriosus, bicuspid aortic valve
- Features in the skin such as: easy bruising, wide scars, soft skin texture, and translucent skin (when it looks almost see-through)
- 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
- Rupture of the spleen or bowel
- Rupture of the uterus during pregnancy
- Instability or malformation of the spine in the neck
- Poor mineralization of the bones (osteoporosis) that can make the bones more likely to break

HOW IS LOEYS-DIETZ DIAGNOSED?

Because LDS was named so recently, not all doctors know about LDS and how to tell if a person has LDS or another connective tissue disorder. A medical geneticist is the kind of doctor most likely to know how to recognize and diagnose LDS. To decide if you have LDS, your doctor will use:

- the health history of you and your family,
- your physical exam,
- the results of special tests including imaging studies of the head, skeleton and blood vessels
- genetic testing to determine if there is a mutation in TGFBR1 or TGFBR2

The special tests should include both an echocardiogram (to study the heart, its valves and the aorta) and 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.

WHAT SHOULD YOU DO IF YOU SUSPECT LOEYS-DIETZ SYNDROME?

Making a correct diagnosis is complicated. People with LDS features need to see a doctor who knows about LDS to decide if they have the disorder.

It is very important that people with LDS get an early and correct diagnosis so they can have the right treatment. (For treatment guidelines see the NMF Fact Sheets on Loeys-Dietz Syndrome medical care.)

HOW CAN YOU LEARN MORE ABOUT LOEYS-DIETZ SYNDROME?

- Call the National Marfan Foundation (NMF) Resource Center at 1-800-862-7326 ext. 26. When you call, you will speak with a nurse who can answer questions and suggest ways to find a doctor who knows about LDS.
- Read the other NMF fact sheets on LDS. They have more information on how LDS is different from Marfan syndrome and LDS management. They are available on-line at www.marfan.org or from the NMF Resource Center.
- Go to the Loeys-Dietz Foundation website at www.loeysdietz.org