



Is it Loeys-Dietz Syndrome or Marfan Syndrome?

Why a Correct Diagnosis Is Important

Doctors who know a lot about Loeys-Dietz syndrome (LDS) believe there are many people diagnosed with Marfan syndrome (MFS) or “atypical” Marfan syndrome who actually have Loeys-Dietz syndrome. Because medical care for LDS is different from care for MFS, it is very important that people have the correct diagnosis. Here are some facts to help you decide if you need to talk with your doctor about LDS.

HOW ARE FEATURES OF LOEYS-DIETZ AND MARFAN SYNDROME ALIKE?

LDS and MFS are both genetic disorders of connective tissue. People with either disorder share many features including:

- Long, thin fingers
- Chest that sinks in (pectus excavatum) or sticks out/pigeon breast (pectus carinatum)
- Curvature of the spine (scoliosis)
- Flexible joints
- Flat feet
- Stretch marks on the skin, not explained by pregnancy or weight gain
- Enlarged or bulging base of the 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)
- Swelling, bulging, or widening of the spinal sac (dural ectasia)

HOW ARE FEATURES OF LOEYS-DIETZ AND MARFAN SYNDROME DIFFERENT?

People with LDS usually do not have the long arms and legs so often seen in people with MFS. Also, people with LDS do not have dislocated lenses in their eyes, whereas, about six out of ten people with MFS have dislocated lenses.

In addition there are several LDS features that set it apart from MFS. These include:

- Arteries that twist and wind (arterial tortuosity)
- Frequent aneurysms and dissections in other parts of the aorta or 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
- 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)
- Premature fusion of the bones of the skull (craniosynostosis)
- Malformation or instability of the spine in the neck
- Collection of fluid in the brain (hydrocephalus)
- Part of the brain (cerebellum) with an abnormal shape (Chiari I malformation)
- Skin issues other than stretch marks including easy bruising, abnormal scars, and a translucent (see-through) quality of the skin that makes it easy to see the veins under the skin
- 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



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- Allergies to both food and things in the environment
- Fragile organs that can cause rupture of the spleen or bowel and rupture of the uterus during pregnancy
- Poor mineralization of the bones (osteoporosis) that can make the bones more likely to break

WHO SHOULD BE CHECKED FOR LOEYS-DIETZ SYNDROME?

People who are in any group listed below should talk with their doctor about the possibility of LDS.

- Those with a diagnosis of MFS or “atypical” MFS who also have any LDS feature (any feature from the second list above).
- Those who have several MFS features but no clear diagnosis, and who have any LDS feature.
- Those with MFS features who have family members who have LDS features.

HOW IS LOEYS-DIETZ SYNDROME DIAGNOSED?

Because LDS was only identified and named in 2005, not all doctors know about LDS and how to tell LDS from MFS. A medical geneticist (a doctor who specializes in genetic disorders) is the kind of doctor most likely to know how to recognize and diagnose LDS. There is genetic testing that can tell if a person has LDS. The genetic testing is looking for mutations in either of the two genes that tell the body how to make proteins called transforming growth factor beta receptor 1 (TGFBR1) and transforming growth factor beta receptor 2 (TGFBR2.) This testing is most helpful when used as part of a complete examination that includes:

- the health history of you and your family
- your physical exam
- the results of special imaging tests including studies of the head, skeleton and blood vessels

WHY IS IT IMPORTANT TO HAVE A CORRECT DIAGNOSIS?

Medical care for LDS is not the same as for MFS. The most important difference is the care of the aorta and other blood vessels. The care is different because life-threatening aneurysms in LDS are more likely to tear and rupture at smaller sizes than in people who have MFS. In LDS, tears and ruptures can also happen at younger ages and in locations not usually seen in MFS. For these reasons, surgery to repair aneurysms is often done earlier in LDS. In addition, imaging tests must look at blood vessels throughout the body using either CT or MR scans from the top of the head to the top of the legs.

There are other LDS medical problems not seen in MFS that need special care. These include cervical spine instability (slipping of the upper part of the spine), congenital heart problems (heart defects at birth), allergies, chronic gastrointestinal problems, rupture of the spleen and bowel, and rupture of the uterus during pregnancy.

LDS medical problems can be managed, but a person needs a correct diagnosis to find the right LDS medical care and counseling.

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 other disorders, how LDS is diagnosed and LDS management. They are available on-line at www.marfan.org or from the NMF Resource Center.
- For laboratories that do LDS genetic testing go to the website of Gene Tests at www.genetests.org and select Laboratory Directory.
- Go to the Loey's-Dietz Foundation website at www.loeydietz.org