

Loeys-Dietz Syndrome

Caring for Cardiovascular Issues

People with Loeys-Dietz syndrome (LDS) need special medical care and management. The most important care involves the cardiovascular system—the heart, the aorta (the main blood vessel that carries blood from the heart) and other blood vessels. Here are some ways to take care of the heart and blood vessels in a person with LDS.

TAKE MEDICATIONS

Medications (drugs) that lower heart rate and/or blood pressure might help prevent bulging or tearing of blood vessels. Doctors often use medications called angiotensin receptor blockers (Losartan, Irbesartan, Telmisartan, etc. and/or beta blockers (Propranolol, Atenolol, Metoprolol, etc) to treat LDS in this way.

There is a preference to avoid the class of blood pressure medications called calcium-channel blockers. In some studies, this medication has been seen more frequently in patients with aortic dissection, and there is some mouse model evidence this worsens ascending aortic aneurysms.

MONITOR YOUR AORTA AND OTHER ARTERIES WITH IMAGING

People with LDS should have:

1. An echocardiogram to check the valves of the heart and the part of the aorta closest to the heart (aortic root) at least once a year. More specialized imaging such as cardiac MRI may be recommended to assess heart size and aortic or mitral valve function.



2. Either a CTA or MRA of the head, neck, chest, abdomen (stomach area) and pelvis (lower stomach area just above the legs) on a regular basis. How often to have these scans depends upon the results of your previous or initial imaging. The scans are looking for aneurysms and/ or dissections (tears) in any of the arteries in the head, neck, chest, abdomen and pelvis. It is recommended that an individual with LDS does not go more than two years without imaging of any portion of the body.

Some people with LDS need to be checked more often than once a year. The frequency of an echocardiogram and other vascular imaging depends on the size of any aneurysms and how fast they are growing. Larger and faster growing aneurysms need more frequent monitoring.

GET REGULAR BUT GENTLE EXERCISE

Most people with LDS can and should be physically active, but should not exercise to the point of becoming exhausted. As a general rule, while exercising, you should be able to comfortably talk with another person without needing to take breaths in the middle of short sentences.

Try walking and gentle hiking, bike riding or swimming as safe ways to stay physically active.

Avoid exercises such as weight lifting, push-ups, chin-ups and sit-ups or other exercises which strain your muscles.

Avoid "contact sports" such as football and basketball or any other activity where there is a high risk of a sharp blow to the head or chest.

Based upon the size of blood vessels or instability of the neck, some people with LDS need to be even more careful and gentle when exercising.

SURGICAL INTERVENTION

Most people with LDS can be helped by a prophylactic or planned cardiovascular or vascular surgery. This means having surgery to remove aneurysms before they cause a life-threatening dissection (tear) or rupture. This is a very important part of LDS care.

The most common vascular surgery is replacement of the aortic root (part of the aorta closest to the heart). Some individuals require replacement of the aortic valve with a bio prosthetic (animal or cadaver) or mechanical valve. Others may be able to keep or spare their native aortic valve, called a valve-sparing aortic root replacement. You should discuss all options with your surgeon, however, the decision about sparing or replacing the valve may ultimately be made in surgery once the surgeon can inspect the valve.

Doctors can use the size of an aneurysm, how fast the aneurysm is growing, where the aneurysm is located, and/or family history of early dissection or rupture to decide when it is time to perform vascular surgery. Your specific gene variant can also help determine need for surgery if we have experience with the variant in other family members or other people reported in the literature.

Vascular surgery is generally very successful in people with LDS. Keep in mind that replacement of the aortic root is also done in people with other connective tissue disorders, especially for Marfan syndrome. However, using Marfan guidelines to decide when to do surgery in a person with LDS can be dangerous because aneurysms in people with LDS can tear or rupture at smaller sizes and at younger ages. Typically, surgery is performed when the aorta crosses the 4.0 cm threshold in LDS types 1, 2, 3 and early-mid 4 centimeters in other types of LDS.

TALK WITH YOUR DOCTOR

LDS is not the same for everyone with the disorder. Some people have more medical problems than others. Therefore, it is very important to talk with your doctor about what care is right for you.

