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Lysosomal Acid Lipase Deficiency

What is lysosomal acid lipase deficiency (LALD)?

Lysosomal acid lipase deficiency (LALD) is a chronic, progressive, inherited disease. Children and adults with LALD do not have a fully functioning enzyme that is needed for the breakdown of certain lipids (fats). As a result of this deficiency, these lipids accumulate in organs (including the liver and spleen) and cells throughout the body causing them to not work properly. This can lead to fatty liver disease, cirrhosis (scarring of the liver) and liver failure.

What causes LALD?

LALD is caused by a deficiency of an enzyme called lysosomal acid lipase (LAL). The LAL enzyme plays a major role in the control of lipid particles known as low density lipoproteins (LDL), a type of cholesterol referred to as "bad cholesterol" which can be linked to cardiovascular disease. Without this enzyme, harmful amounts of various lipid particles build up in a part of the cell known as the lysosome. This fat accumulation, can lead to significant health consequences including damage to the liver and other organs.

LALD in infants

Very low levels of the LAL enzyme lead to early onset LALD, also known as Wolman disease (WD). It is estimated that LALD occurs in about 8 live births per year. However, the rate may be higher if the parents are of Persian-Jewish ancestry. LALD presents in infants as a rapidly progressive disease with life-threatening complications occurring most frequently within the first six months of life. Infants with this disease develop liver failure and cannot grow properly because they have difficulty absorbing nutrients from food. Affected infants generally do not survive past one year of age.

LALD in children and adults

Late onset LALD is also known as cholesteryl ester storage disease (CESD) and can affect children and adults. It is estimated that the prevalence of CESD is 1 in 40,000 individuals. The deficiency of the LAL enzyme leads to a build-up of fat in the liver, spleen and other parts of the body. In children and adults, the disease may not be easily recognized because individuals may feel well and have no symptoms. Many lipids accumulate in the liver, and as a result, the liver becomes dysfunctional and may become enlarged. Ultimately, scarring and liver failure may occur. In addition, high levels of lipids in the blood can lead to cardiovascular complications, such as coronary artery disease (CAD) and stroke.

How is LALD diagnosed?

Because some of the signs and symptoms associated with LALD are similar to those of other more common disorders, it may take months or even years for people with LALD to get an accurate diagnosis. Also, since the initial signs and symptoms may seem rather mild, patients and healthcare providers may not suspect a progressive disease for a long time.

An accurate and definitive diagnosis of LALD can be made with a blood test that measures the activity of the LAL enzyme. LALD can also be diagnosed through genetic testing. You need to consult your healthcare professional regarding testing for LALD.

What are the symptoms of LALD?

Infants, children and adults who suffer from LALD experience a range of serious health problems. The lack of the LAL enzyme can lead to a build-up of fatty material in a number of body organs including the liver, spleen, gut, in the wall of blood vessels and other organs. Many signs of LALD may not be visible and can only be determined through blood tests and examination by a healthcare provider. However, some common symptoms and signs of LALD in infants may include:

- Failure to grow
- Difficulty absorbing nutrients from food (malabsorption), presence of fat in feces
- Persistent diarrhea and vomiting
- Swollen belly
- Jaundice (yellowing of the skin and eyes).

The signs and symptoms of LALD in children and adults may not be present at all. As the liver damage progresses, symptoms may include:

- Enlarged abdomen from fluid accumulation
- Easy bruising or bleeding
- Jaundice (yellowing of the skin and eyes).

What treatments are available for LALD?

Currently there are no approved treatments for LALD. Medication to control cholesterol levels is recommended in order to prevent the early development of fat deposits in blood vessels. Use of enzyme replacement therapy with recombinant human LAL in short-term studies has shown some promise but this is still under investigation. Please discuss treatment options with your healthcare provider.

This information is current for September 2015.