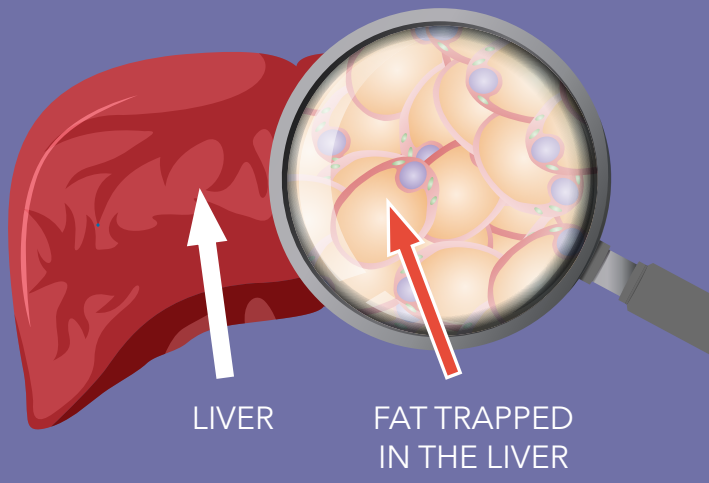
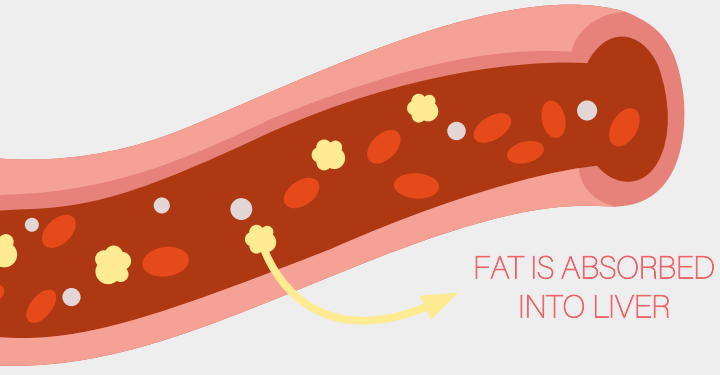
Lysosomal Acid Lipase Deficiency (LAL-D)

FOUNDATION
of the National Lipid Association

Quick Facts

- Individuals with LAL-D lack an enzyme necessary to properly digest fat in the liver
- LAL-D is inherited genetically (autosomal recessive)
- You could be a carrier even though you don't have the disorder yourself
- There is no cure yet, but there are treatment options
- If LAL-D is not recognized early and treated effectively, most infants with LAL-D do not live longer than one year





Why do we need Lysosomal Acid Lipase?

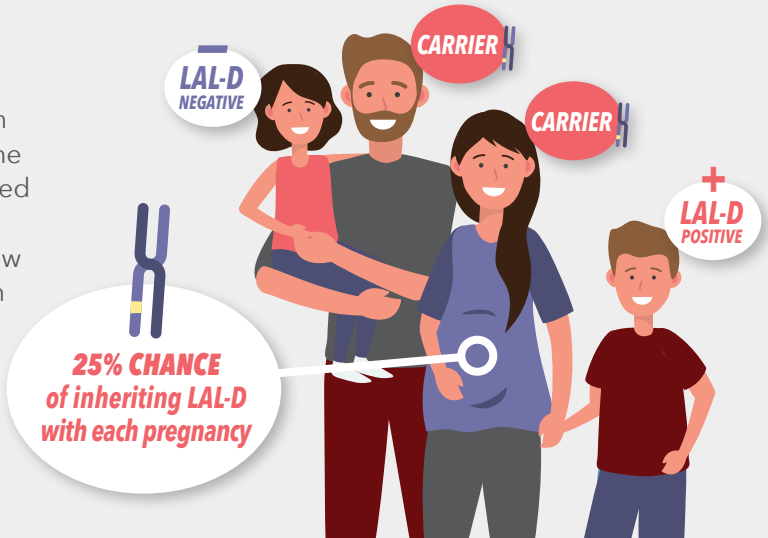
Lysosomal acid lipase is an enzyme in the liver that helps the body digest fat. The fat normally found in the blood is either made by our bodies or taken from the food we eat. This fat promotes good health and helps children achieve normal growth and development. To be useful to our body, the fat in the blood is absorbed into the liver where it is digested.

What is Lysosomal Acid Lipase Deficiency?

This condition, also referred to as "LAL-D", is a genetic (inherited) disorder that affects infants, children, and adults. In the absence of the lysosomal acid lipase enzyme, fat cannot be digested, causing it to become trapped in the liver. As this fat accumulates, it causes the level of fat in the blood to increase and can damage the liver and other important organs.

How is LAL-D transmitted?

LAL-D is an autosomal recessive genetic disorder. Although neither the child's mother nor father is affected by LAL-D, the condition is passed down when the child receives an affected copy of the gene causing LAL-D from both parents. Genes help determine everything from the color of our eyes to how our body's work. The risk of having a child with LAL-D when both parents are carriers of the LAL-D gene is 25%, each time the mother becomes pregnant.



How can I tell if I or my child have Lysosomal Acid Lipase Deficiency?

A blood test can tell whether you are deficient of lysosomal acid lipase. For those with an abnormal test, genetic testing is often recommended to 1) confirm the diagnosis of Lysosomal Acid Lipase Deficiency; 2) determine the risk or severity of disease; 3) help you and your health care providers decide on the best treatment plan; and 4) provide valuable information to identify the likelihood of passing the LAL-D gene on to children in the future.

Infants who have LAL-D often have symptoms in the first several days or weeks of life. Sadly, if the LAL-D is not recognized early and treated effectively, most infants with LAL-D generally do not live longer than one year.

If not present during the first year of life, symptoms of LAL-D usually occur before 20 years-of-age, although the first symptoms may occur as an adult. Rather than the severe symptoms seen during the first year of life, most with LAL-D may have symptoms that go unrecognized.

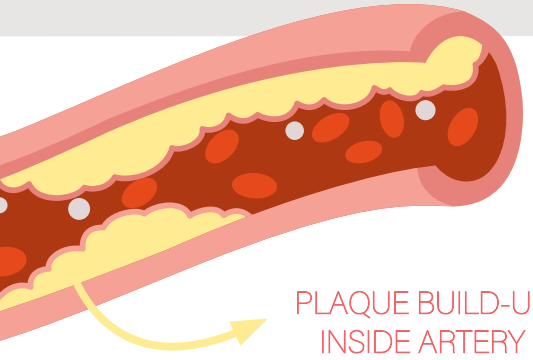
Symptoms in infants include:

1. Poor feeding
2. Diarrhea
3. Enlargement of the liver
4. Inability to gain weight properly (a condition often referred to as "failure to thrive")

Symptoms in children and adults include:

1. High levels of LDL-cholesterol (often referred to as "bad cholesterol")
2. Liver problems

LAL-D is often suspected because of abnormal blood test results. Although children and adults with LAL-D are not as severely affected as infants with the condition, they do experience multiple health problems and in general, do not live as long as healthy peers.



What health problems can be caused by Lysosomal Acid Lipase Deficiency?

Health problems related to LAL-D in children and adults, including liver failure, tend to occur over years. Due to the severe and lifelong elevation of "bad cholesterol," children and adults with LAL-D may develop clogged arteries, ultimately causing damage by reducing blood flow to organs, including the heart, brain, and kidneys. This damage can result in a heart attack or stroke at a very early age (e.g., 40-50 years of age or younger). Accumulation of fat in other organs may also contribute to poor health in affected individuals.

Is LAL-D curable?

LAL-D is not curable, but fortunately, we now have an enzyme to replace the one that is deficient in those affected by LAL-D. If diagnosed and treated early, infants with the severe form as well as children and adults with LAL-D have significantly improved health and survival. Since the treatment is relatively new, it is hoped that the health benefits seen with treatment will continue as individuals with LAL-D grow older. Those whose "bad cholesterol" level remains high after starting enzyme replacement therapy may benefit from medication that can reduce their LDL cholesterol to a safer level (<70 mg/dL).

What should children and adults with LAL-D do to stay healthy?

- Eat a heart-healthy diet
- Participate in 30-60 minutes of moderate-to-vigorous physical activity every day
- Maintain a healthy weight
- Avoid the use of tobacco/nicotine

While helpful in those with LAL-D, maintaining heart healthy habits are important but generally not enough to stay healthy in the absence of enzyme replacement therapy or other forms of treatment.