

Lysosomal Acid Lipase Deficiency

This condition is sometimes called:

- Acid Cholesteryl Ester Hydrolase Deficiency, Type 2
- Acid Lipase Disease
- Cholesteryl Ester Storage Disease (CESD)
- Cholesterol Ester Hydrolase Deficiency
- Early-onset LAL Deficiency (Wolman Disease)
- LAL Deficiency
- Late-onset Lysosomal Acid Lipase Deficiency (CESD)
- Wolman Disease (early onset LAL Deficiency)

Related Disorders may include:

- Non-Alcoholic Fatty Liver Disease (NAFLD)
- Non-Alcoholic Steatohepatitis (NASH)
- Alcoholic Liver Disease
- Cryptogenic Cirrhosis
- Niemann-Pick Disease (NPD) Type C
- Chanarin Dorfman Syndrome

LAL Deficiency

Lysosomal Acid Lipase (LAL) Deficiency belongs to a family of diseases called Lysosomal Storage Disorders. These are genetic conditions, and like some other genetic disease (eg cystic fibrosis) parents may not know they are carriers of the abnormal gene until they have an affected child. If both parents are carriers for LAL Deficiency, there is a one in four chance that their baby will inherit the gene that prevents the body from producing lysosomal acid lipase (LAL). The LAL enzyme breaks down fatty material (cholesteryl esters and triglycerides), and the lack of the LAL enzyme results in a build-up of these materials in the liver, the gut or other important organs including the walls of blood vessels.

Early-Onset and Late-Onset Lysosomal Acid Lipase (LAL) Deficiency

The early-onset form of LAL Deficiency, sometimes called Wolman Disease, strikes 1-2 babies for every million births and is rapidly fatal, usually in the first year. There are signs of liver damage in the early-onset babies, but the cause of death is a result of growth failure. LAL Deficiency manifests more frequently in children, adolescents or adults than it does infants. Late-onset LAL Deficiency, sometimes called Cholesteryl Ester Storage Disease (CESD), affects 25 individuals per million births and may lead to cirrhosis of the liver, liver failure and death. There also appears to be an increased risk of strokes because of potential build-up of lipid in the walls of major arteries (atherosclerosis).

Many of these signs and symptoms of LAL Deficiency are experienced by patients with other, more common liver conditions such as Non Alcoholic Fatty Liver Disease (NAFLD), Non Alcoholic Steatohepatitis (NASH), Alcoholic Liver Disease or Cryptogenic Cirrhosis. LAL Deficiency and these other conditions share a common trait of liver damage. It is likely that patients who suffer from the genetic condition of LAL Deficiency go unnoticed and undiagnosed because some of their signs and symptoms resemble those of this larger population.

There is no treatment approved for LAL Deficiency. At present, medical care focuses on managing the symptoms. Intravenous nutritional support is sometimes used for early-onset LAL Deficiency if bone marrow transplant is being considered. For late-onset LAL Deficiency, combining drugs that reduce blood cholesterol with a low cholesterol diet has been effective at reducing some of the symptoms.

Synageva BioPharma, however, has launched a clinical program to investigate a human recombinant lysosomal acid lipase as an enzyme replacement therapy for LAL Deficiency. Enzyme replacement therapies have been successful in treating other lysosomal storage disorders such as Gaucher Disease, Fabry Disease and Pompe Disease. For more information on Synageva's clinical trials, go to [Synageva clinical trials](#) or [EU Clinical Trials Register](#), use "Synageva" or "Lysosomal Acid Lipase" as search terms).