

Alpha-1 Antitrypsin Deficiency

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About the Liver

- The human liver is an organ and gland in the human body.
- The liver is an essential organ, performing hundreds of functions necessary to sustain life.
- Weighing, on average, about three pounds in an adult, the liver is the largest internal organ.
- The liver is located on the **right side of the body**, under the ribs.
- The liver has hundreds of jobs but the main job is to control most chemical levels in the blood.
- The liver has two main parts: the larger right lobe and the smaller left lobe.

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What is Alpha-1 Antitrypsin Deficiency

Alpha-1 Antitrypsin Deficiency is a genetic condition that can cause damage to the lungs and/or the liver.

There are about 100,000 people in the United States living with Alpha-1. Worldwide, the number of people with Alpha-1 is 1 in every **1,500 to 3,500 people of European ancestry.** People of any racial or ethnic group can get Alpha-1, but it is more common in people of northern European Backgrounds. There is no cure for Alpha-1, but treatment can help people with Alpha-1 manage symptoms. Avoiding certain behaviors, like smoking and drinking, can help slow the progression of Alpha-1 lung disease.

The low level of AAT in the blood occurs because the AAT is abnormal and cannot be released from the liver at the normal rate. This leads to a buildup of abnormal AAT in the liver that can cause liver disease. Though Alpha-1 is diagnosed by a blood test, other tests may be performed to determine the severity of a person's Alpha-1. It is a progressive condition, which means it may worsen over time. There is no cure for Alpha-1, but some symptoms can be managed with treatment.

Treatment and Therapy Options

Augmentation therapy is a long-term treatment that will increase the levels of AAT in your blood by giving you donated AAT. It cannot reverse lung damage that has already occurred, but it may help to slow lung damage. Augmentation therapy generally requires a weekly infusion and is reserved for those individuals with the lowest AAT levels. If AAT deficiency affects your liver, you should avoid drinking alcohol.

www.lung.org

Did you know that Gene Mutations Causes Alpha-1?

The alpha-1 antitrypsin (AAT) is abnormal due to a defective gene and cannot be properly released from the liver into the bloodstream. This leads to a build-up of abnormal AAT in the

liver that can cause liver disease. Alpha-1-antitrypsin
deficiency is inherited in an autosomal
codominant manner, which means each person
inherits one copy of the gene from each parent,
and both copies affect the amount of alpha-1-antitrypsin
produced. AATD is caused by mutations in the
SERPINA1 gene, which leads to low levels and/or abnormal
forms of the AAT protein.Alpha-1 antitrypsin deficiency (AATD)
is caused by mutations in the SERPINA1 gene, which
encodes the alpha-1 antitrypsin (AAT) protein.

alpha1mz.org

Alpha-1 Antitrypsin Deficiency Policies

The John W. Walsh Alpha-1 Home Infusion Act of 2023

This bill provides for Medicare coverage of treatment for alpha-1 antitrypsin (AAT) deficiency (a protein deficiency that raises the risk of lung and other diseases). Specifically, the bill provides for coverage of at-home augmentation therapy for beneficiaries with emphysema as a result of severe hereditary AAT deficiency. Treatment must be provided through qualified home infusion therapy suppliers; beneficiaries must be under the care of a physician, nurse practitioner, or physician assistant

Alpha1.org Congress.gov