

# Alpha-1- Antitrypsin Deficiency



## What is alpha-1-antitrypsin deficiency ?

**A**lpha-1-antitrypsin (A1AT) deficiency is an inherited (genetic) disease in which a protein known as A1AT is unable to be released from the liver into the blood in adequate amounts.

This causes a deficiency of the A1AT protein in the circulation. A1AT is a specialized protein that blocks the action of other proteins important in inflammation (swelling) and breakdown of tissues in the body. When the A1AT protein is deficient in the circulation, tissue inflammation and tissue damage is more severe, especially in the lungs. The defective A1AT protein that remains in the liver may cause liver inflammation (hepatitis) that can progress to cirrhosis (permanent liver scarring) and liver failure. A1AT deficiency is quite common, with about 1 in 1500 to 2000 people affected. Both parents must be carriers of the genetic defect to have a child with A1AT deficiency. Although "carrier" parents partially produce the abnormal A1AT protein, they usually have no, or very minimal symptoms and they may not realize they are partially affected until their child is diagnosed with the condition.

## What are the symptoms?

About 10% of newborns with liver disease will be diagnosed with A1AT deficiency. Newborns may develop jaundice (yellowing of the eyes and skin) as part of inflammation of the liver associated with A1AT deficiency. Older children and teens can present with a liver that has been inflamed for a long time, causing scarring (cirrhosis) to develop. Lung disease usually develops only in adulthood. Adults with A1AT deficiency may have difficulties with COPD (chronic obstructive pulmonary disease), emphysema, chronic bronchitis, asthma, coughing, and repeated lung infections.

## How is it diagnosed?

Testing is usually done when a child has liver inflammation of uncertain cause, or when a family history suggests A1AT liver disease. The standard screening test is to measure the amount of A1AT protein in the circulation. In cases of A1AT deficiency, the serum A1AT level will be low. When a low A1AT level is found, an additional test is usually done to identify the types of abnormal A1AT proteins that are in the circulation. In rare cases, a more detailed analysis of the DNA in the gene may be necessary.

The amount of liver swelling and damage can be assessed by (a) ultrasonography of the abdomen (sonogram), b) blood tests of liver inflammation and function, and (c) a liver biopsy where a small piece of liver tissue is obtained and then examined under the microscope to search for changes typical of A1AT deficiency.

## How is it treated?

There is no cure for A1AT deficiency. Controlling symptoms and attempting to prevent the development of complications are the mainstays of therapy for A1AT deficiency. It is very important for the affected person and those around him/her never to smoke in order to protect their lungs. Vaccination should be given to protect against viruses that can further damage the liver, such as Hepatitis A and Hepatitis B. It is difficult to predict how severely a liver will be affected. Liver function tests are followed over time. Some patients may have minimal liver disease during their entire lifetime; however a small number will eventually require a liver transplant. It is hoped that in the near future, with advances in gene therapy, it will be possible to supplement the missing protein or correct the gene defect.

For more information or to locate a pediatric gastroenterologist in your area please visit our website at: [www.naspghan.org](http://www.naspghan.org)

**IMPORTANT REMINDER:** This information from the North American Society for Pediatric Gastroenterology, Hepatology and Nutrition (NASPGHAN) is intended only to provide general information and not as a definitive basis for diagnosis or treatment in any particular case. It is very important that you consult your doctor about your specific condition.

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