

# Alpha-1 Antitrypsin Deficiency

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## KEY POINTS

- Alpha-1 antitrypsin deficiency (AATD) is an inherited life-long condition caused by a lack of AAT proteins in the blood. AAT proteins protect the lungs and liver from damage. AATD may lead to emphysema, which is a serious disease that gradually destroys the lungs and makes it hard to breathe, or to cirrhosis, which is scarring of the liver.
  - The most important part of treatment is to quit smoking or vaping. Treatment usually includes medicines and may include a lung or liver transplant.
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## What is alpha-1 antitrypsin deficiency?

Alpha-1 antitrypsin deficiency (AATD) is a life-long condition caused by a lack of AAT proteins in the blood. AAT proteins protect the lungs and liver from damage. AATD may lead to emphysema, which is a serious disease that gradually destroys the lungs and makes it hard to breathe, or to cirrhosis, which is scarring of the liver.

## What is the cause?

Alpha-1 antitrypsin deficiency is inherited. Genes are in each cell of the body. They contain the information that tells the body how to develop and work. Changes in the genes can be passed from parents to children.

Exposure to tobacco smoke, chemicals, and dust can worsen AATD.

## What are the symptoms?

Symptoms may include:

- Shortness of breath after mild activity
- Wheezing
- Feeling very tired
- Unplanned weight loss
- Frequent lung infections

If AATD affects the liver, symptoms may include:

- Yellowing of skin or eyes
- Swelling in the abdomen
- Itching all over
- Nausea and vomiting
- Clay colored bowel movements
- Darker than normal urine

## How is it diagnosed?

Your healthcare provider will ask about your symptoms and medical history and examine you. A blood sample or a swab of cells from the inside of the cheek are sent to the lab to look for the gene changes that cause AATD. Other tests may include:

- Chest X-rays
- Spirometry, which is a breathing test. You will breathe into a tube to measure how well air moves into and out of your lungs. The test can show how well your lungs are working.
- CT scan, which uses X-rays and a computer to show detailed pictures of your lungs and liver

## How is it treated?

If you are a smoker or vaper, the most important part of your treatment is to quit smoking or vaping. Stopping smoking or vaping may help improve your symptoms and slow the damage to your lungs, depending on the extent of your emphysema. Talk to your healthcare provider about ways to quit smoking. You might find it helpful to join a quit-smoking program, use nicotine patches or gum, or try one of the prescription medicines that can help you quit. These methods work best when they are used together and with the guidance of a trained counselor or healthcare provider.

Your healthcare provider may prescribe the following medicines:

- Bronchodilator: This medicine relaxes the muscles of the airways. It makes the openings inside the airways bigger, so it's easier to breathe. Usually you take this medicine by inhaling it (breathing it into your lungs), but it may also be taken as pills or liquid.
- IV infusion of medicines to replace AAT protein
- Steroid: This medicine helps keep down the swelling inside your airways. You may need steroids if your symptoms are severe or if you start having symptoms more often. Steroids may be inhaled or taken as a pill.

Using a steroid for a long time can have serious side effects. Take steroid medicine exactly as your healthcare provider prescribes. Don't take more or less of it than prescribed by your provider and don't take it longer than prescribed. Don't stop taking a steroid without your provider's approval. You may have to lower your dosage slowly before stopping it.

A lung or liver transplant may be an option. Talk with your healthcare provider about this.

## How can I take care of myself?

Follow the full course of treatment prescribed by your healthcare provider. In addition:

- Don't smoke or vape.
- Avoid exposure to secondhand smoke, dust, polluted air, fumes, chemicals, and mold.

- Get a flu shot every year and avoid close contact with people who have colds or the flu.
- Wash your hands often and well, especially when you are in public places or around children.
- Eat a variety of healthy foods and maintain a healthy weight.
- Stay physically active as advised by your provider.
- If you want to drink alcohol, ask your healthcare provider how much is safe for you to drink.
- Ask about getting pneumococcal and hepatitis vaccines to protect against a serious infection.

Ask your provider:

- How and when you will get your test results
- If there are activities you should avoid and when you can return to normal activities
- How to take care of yourself at home
- What symptoms or problems you should watch for and what to do if you have them

Make sure you know when you should come back for a checkup.

## **How can I help prevent AATD?**

People who have the gene may pass the disease on to their children. Couples can get tested for the AATD gene. If one of you has the gene, you will need to decide whether to take the chance of passing the disease on to your children. You may want to talk with a genetic counselor if anyone in your family has AATD.

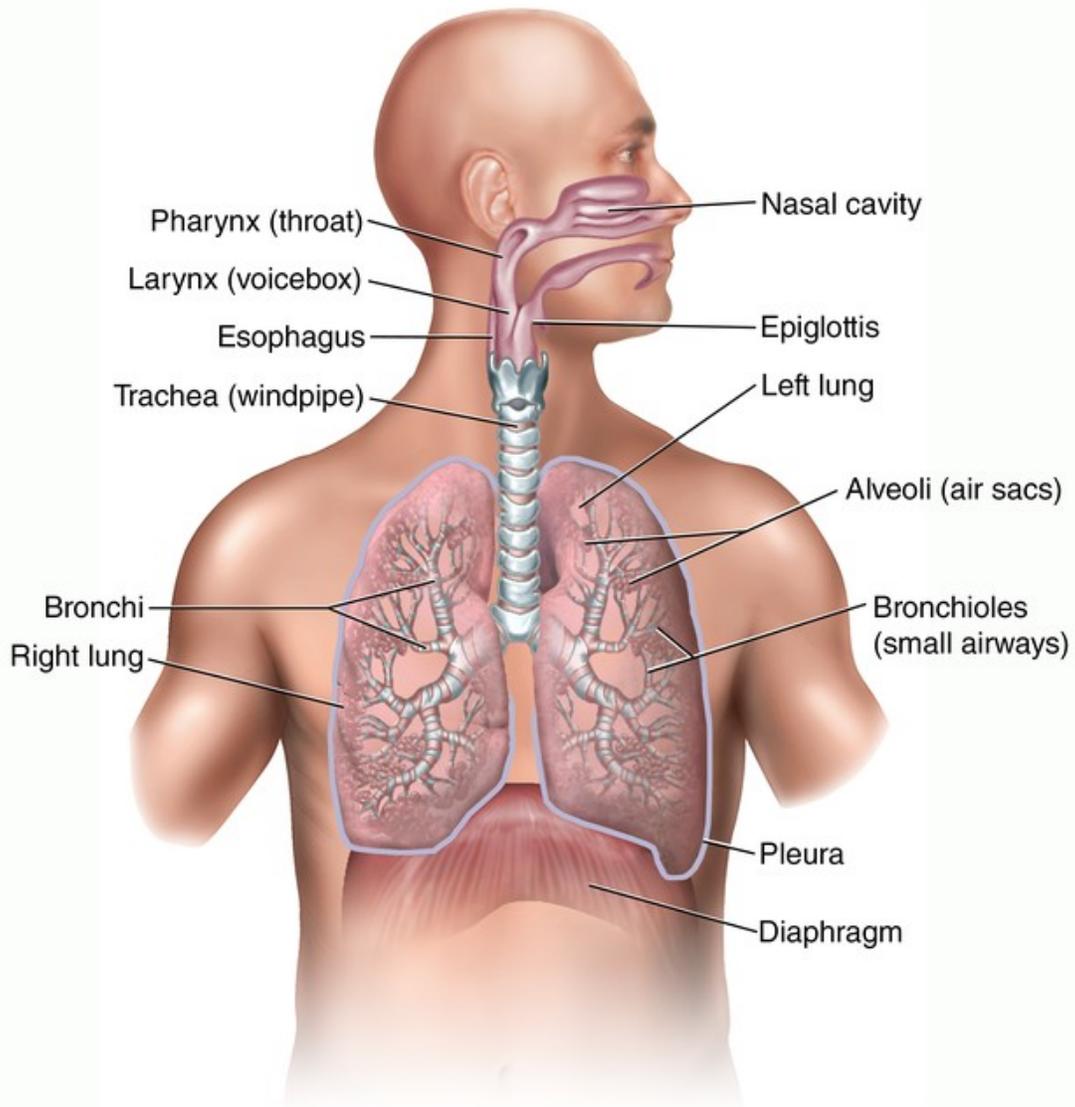
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# Respiratory System



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