

What is Alpha-1 Antitrypsin Deficiency?

Alpha-1 antitrypsin deficiency is an inherited form of emphysema (em-fuh-ZEE-muh). People with the condition, also known as AAT Deficiency or alpha-1, do not have enough of a protein called alpha-1 antitrypsin (AAT) in their blood. This protein is made in the liver, and it protects the lungs so they can work normally. Without enough AAT, the lungs can become damaged by emphysema. Alpha-1 also can also cause liver damage.



What is emphysema?

Emphysema is a condition that involves damage to the walls of the air sacs (*alveoli*) of the lung. Normally there are more than 300 million alveoli in the lung. These alveoli are stretchy and springy, like little balloons. Like a balloon, it takes effort to blow up normal alveoli; however, it takes no energy to empty the air sacs because they spring back to their original size.

In emphysema, the walls of some of the alveoli have been damaged. When this happens, the alveoli lose their stretchiness and trap air. Since it is difficult to push all of the air out of the lungs, the lungs do not empty easily and therefore contain more air than normal. This is called air trapping and causes hyperinflation in the lungs. In alpha-1, emphysema occurs more in the lower parts of the lungs than the upper parts. The combination

of constantly having extra air in the lungs and the extra effort needed to breathe causes a person to feel short of breath. Airway obstruction occurs in emphysema because the alveoli that normally support the airways open cannot do so during inhalation or exhalation. Without their support, the breathing tubes collapse, causing blockage (obstruction) to the flow of air.

What causes Alpha-1?

Alpha-1 is an inherited condition. Every person inherits two AAT genes—one from each parent. Inheriting two abnormal AAT genes causes alpha-1 deficiency. A person who inherits only one abnormal gene is an alpha-1 “carrier.” While a carrier’s AAT levels may be lower than normal, the risk of major health problems is much less than in a person with two abnormal genes.

Does everyone with two abnormal Alpha-1 genes develop disease?

Not everyone who inherits two abnormal AAT genes gets emphysema and/or liver disease. Some people never have symptoms. Some have only mild symptoms. Other people can have severe lung problems, liver problems, or both. Smoking is known to make lung disease worse if you have alpha-1.

What are the signs and symptoms of Alpha-1?

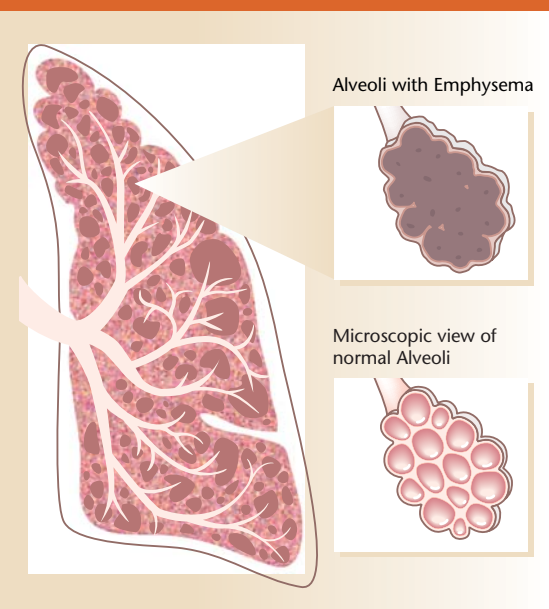
Alpha-1 lung symptoms are most likely to appear after age 30, but may emerge much later. The first symptom is usually shortness of breath during daily activities. Other symptoms include wheezing and decreased ability to exercise.

How is Alpha-1 diagnosed?

Alpha-1 can be detected through blood tests or a new test that painlessly takes a sample of DNA from the cells inside your mouth. One type of blood test measures the body’s level of AAT. If the AAT level is lower than normal, your

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healthcare provider may order a *genotype* or a *phenotype* blood test. These genetic analysis tests look at the amount and type of AAT being produced and compare it with normal patterns.

If you are diagnosed with alpha-1, talk to your doctor about having other family members, including your siblings and children, tested for alpha-1 deficiency.

Alpha-1 testing is recommended for certain groups of people, including those who have:

- Family history of alpha-1
- Early-onset emphysema (less than 45 years old)
- Emphysema without an obvious risk factor such as smoking or occupational exposure to a substance known to cause the disease
- Emphysema that is worse at the bottom of the lungs
- Chronic asthma (in adolescents and adults)
- Recurrent pneumonia or bronchitis
- Unexplained liver disease

The doctor also may recommend tests including a chest X-ray, a lung function test (a breathing test to find out how your lungs function compared with people with normal lungs), an arterial blood gas (measuring the level of oxygen in the blood) and liver function tests.

What are the treatments for Alpha-1?

At this time, there is no cure for Alpha-1, but there are treatments that can improve symptoms. Your healthcare provider may prescribe medications such as bronchodilators or inhaled steroids to help open your airways. Your healthcare provider also may recommend pulmonary rehabilitation to improve your breathing. (For additional information on pulmonary rehabilitation, see ATS Fact Sheet: Questions About Pulmonary Rehabilitation at <http://www.thoracic.org/sections/education/index.html>) Patients with severe alpha-1 may be candidates for a lung transplant. A treatment called *augmentation therapy*, which may slow down or stop the destruction of lung tissue, may also be prescribed. This treatment increases the level of AAT in the blood. It is given intravenously (through a vein), and is usually given once a week for life.

Can I protect myself from lung damage if I have Alpha-1?

If you have been diagnosed with alpha-1, one of the most important things you can do is to **quit smoking**, and protect yourself from secondhand smoke. Tobacco smoke irritates and damages the lungs. In people with alpha-1, inhaling tobacco smoke speeds up the damage to the lungs.

There are a number of other steps you can take to reduce the risk of breathing problems if you have alpha-1. These include:

- Stay indoors when air quality is poor. Keep windows closed.

- Avoid dust whenever you can. If you have to clean, wear a mask, particularly when shaking rugs, vacuuming, sweeping, and dusting.
- People with alpha-1 may be more severely affected by respiratory infections. To reduce the risk of developing serious complications from these infections, get flu and pneumonia vaccines and avoid people who are ill.
- Wash your hands frequently—it's the best way to avoid catching a cold or the flu.
- Contact your healthcare provider at the first sign of a cold or other lung problem so you can try to keep it from getting worse.
- Exercise regularly to stay in shape.
- Avoid excessive alcohol—drinking may increase the risk of developing liver problems in people with Alpha-1.
- Ask your doctor whether you would benefit from AAT augmentation therapy.

Source: ATS/ERS Statement: Standards for the Diagnosis and Management of Individuals with Alpha-1 Antitrypsin Deficiency, <http://www.thoracic.org/sections/publications/statements/pages/respiratory-disease-adults/alpha1.html>

Additional Lung Health Information

American Thoracic Society

<http://www.thoracic.org/sections/publications/statements/index.html> and

<http://www.thoracic.org/sections/education/patient-education/index.html>

Alpha-1 Foundation

<http://www.alphaone.org>

American Lung Association

<http://www.lungusa.org>

Rx Key Points

- ✓ Quit smoking and stay away from second-hand smoke
- ✓ Exercise regularly to stay in shape
- ✓ Avoid exposure to outdoor and indoor pollution like ozone, dust and fumes
- ✓ Get your flu and pneumonia vaccines as recommended by your health care provider
- ✓ Contact your healthcare provider early if you have a cold or other respiratory illness
- ✓ Avoid excessive alcohol consumption
- ✓ Get regular health check-ups and lung function testing

Doctor's Office Telephone: