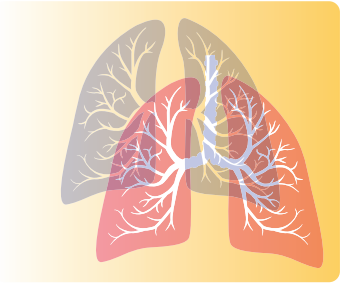
What Is Alpha-1 Antitrypsin Deficiency?

Alpha-1 antitrypsin deficiency is a genetic condition that decreases lung protection resulting in an inherited form of emphysema (em-fuh-ZEE-muh). People with the condition, also known as AAT Deficiency or alpha-1 antitrypsin deficiency, 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.

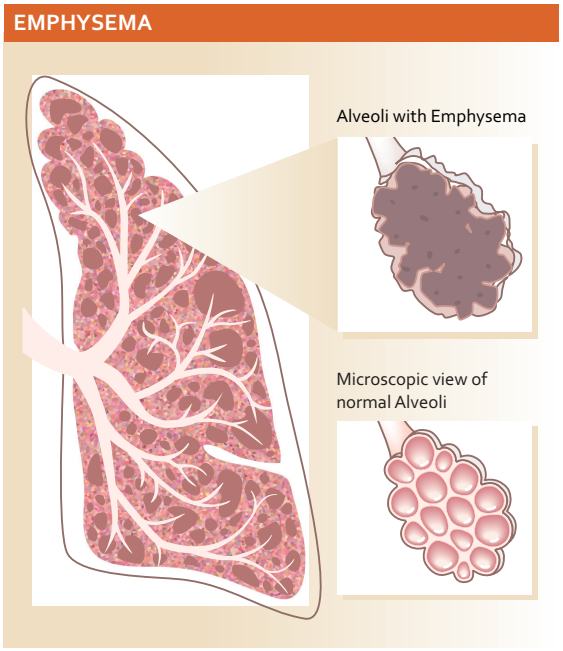


Without enough AAT, the lungs can become damaged by smoke, dust and fumes. AAT deficiency also can also cause liver damage.

What is emphysema?

Emphysema is a condition that involves damage to the walls of the air sacs (alveoli) of the lung producing holes in 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. The lungs do not empty easily because air is trapped in the holes. This is called air trapping and causes hyperinflation (lung gets larger than normal). In AAT deficiency, 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 blockage (obstruction) occurs in emphysema because the alveoli that normally help keep the airways open cannot do so during inhalation or exhalation. Without their support, the breathing tubes collapse, causing blockage to the flow of air.



What causes AAT deficiency?

Alpha-one antitrypsin deficiency is an inherited condition. Every person inherits two AAT genes—one from each parent. Inheriting two abnormal AAT genes causes very low levels of AAT in the blood. A person who inherits only one abnormal gene is an AAT "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 AAT deficiency 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 AAT deficiency.

What are the signs and symptoms of AAT deficiency?

AAT deficiency lung symptoms usually appear after age 30, but may emerge earlier or much later. The first symptom is usually shortness of breath during daily activities. Other symptoms include wheezing and decreased ability to exercise. Some people will have excess mucus that they cough up. This can occur if they also have airway damage called bronchiectasis (BRONK-e-EK-ta-sis) from recurrent infection. For more information on bronchiectasis, see ATS Patient Information Fact sheets on Bronchiectasis.

How is AAT deficiency diagnosed?

AAT deficiency only can be detected through blood tests. One type of blood test measures the body's level of AAT. If the AAT level is lower than normal, your 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. One test in a lifetime is sufficient for diagnosis.

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AAT deficiency testing is recommended for certain groups of people, including those who have these at risk conditions:

- Family history of AAT deficiency
- Early-onset pulmonary emphysema
- Everyone who has chronic obstructive pulmonary disease (COPD)
- Unexplained liver disease
- Asthma in which lung function fails to return to normal with therapy
- Adults with bronchiectasis without evident cause
- Shortness of breath and cough in multiple family members

The healthcare provider 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. A chest computed tomography (CT) scan is often used to determine the extent of emphysema and/or bronchiectasis present.

What are the treatments for AAT deficiency?

At this time, there is no cure for AAT deficiency, 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. People who have severe emphysema from AAT deficiency may be candidates for a lung transplant. A treatment called AAT 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. For more information on any of these treatments, see ATS Patient Information Series fact sheets on COPD, lung transplantation, and pulmonary rehabilitation.

Can I protect myself from lung damage if I have AAT deficiency?

If you have been diagnosed with AAT deficiency, one of the most important things you can do is to stop smoking, and protect yourself from secondhand smoke, dust and fume exposure. Tobacco smoke irritates and damages the lungs and speeds up lung damage in AAT. For help with stopping smoking, see ATS Patient Information Series fact sheets on Tobacco.

Other things you can do to reduce the risk of problems with AAT deficiency. These include:

- Stay indoors when air quality is poor such as with an ozone pollution alert. Keep windows closed.
- Avoid dust whenever you can. If you have to clean, wear a mask, particularly when shaking rugs, vacuuming, sweeping, and dusting.
- Avoid any job or occupation where there is dust exposure (See ATS Patient information Series fact sheet on *Occupational Lung Disease*).
- People with AAT deficiency may be more severely affected by respiratory infections. You should get pneumococcal pneumonia vaccines and a yearly influenza vaccine (flu shot). Avoid contact with people who are ill when possible.
- 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 AAT deficiency.
- Monitor liver and lung function at least yearly with a healthcare provider knowledgeable about AAT deficiency.
- Ask your healthcare provider whether you would benefit from AAT augmentation therapy.
- Consider joining a support group, participating in ATS/PAR Lung Disease Week on Alpha-1, or attending an Alpha-1 Foundation Education day.

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*deceased

Rx *Action Steps*

- ✔ 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 healthcare provider
- ✔ Contact your healthcare provider early if you have a cold or other respiratory illness
- ✔ Avoid excessive alcohol consumption
- ✔ Get regular health check-ups with lung and liver function testing
- ✔ Consider joining a support group or attending a disease specific education day

Healthcare Provider's Contact Number:

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Resources

American Thoracic Society
http://www.thoracic.org

- Bronchiectasis
- COPD
- Lung Function Testing
- Occupational Lung Disease
- Pulmonary Rehabilitation
- Tobacco

Alpha-1 Foundation
- http://www.alphaone.org