



Understanding Alpha-1 Antitrypsin Deficiency

Your health care team offers this information to answer your questions about alpha-1 antitrypsin deficiency. It explains the signs and symptoms of the disorder as well as risk factors and treatment. When you understand more about alpha-1 antitrypsin deficiency, you will be able to play a more active role in your care.

What is alpha-1 antitrypsin deficiency?

Alpha-1 antitrypsin is a protein that is made in the liver. The liver releases this protein into the bloodstream.

Alpha-1 antitrypsin protects the lungs so they can work normally. Without enough alpha-1 anti-trypsin, the lungs can be damaged, and this damage may make breathing difficult.

Alpha-1 antitrypsin deficiency is an inherited (passed down from parents) disorder that causes low levels of, or no alpha-1 antitrypsin in the blood.

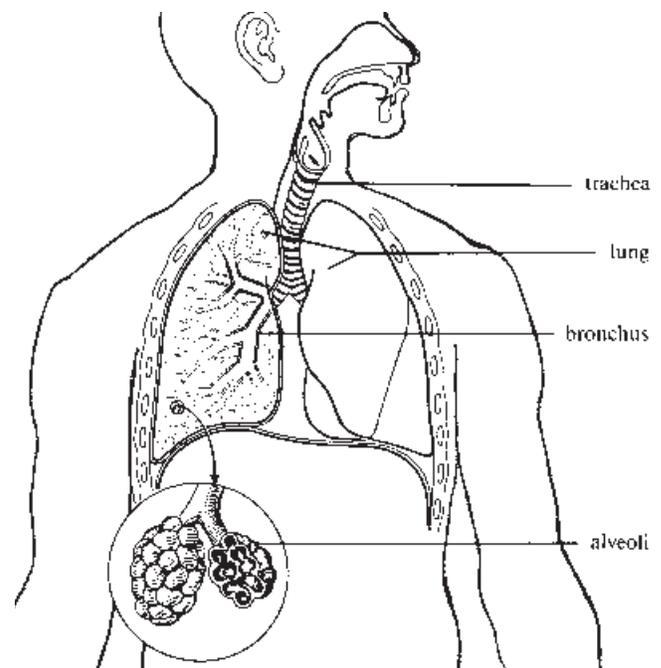
How do normal lungs work?

Air usually enters the nose and mouth and goes down the air tube (trachea) to two main air passages (bronchi). These passages allow air to go into the right and left lung.

Each bronchus branches out into grape-like air sacs called alveoli. Through the alveoli, oxygen enters the bloodstream during breathing in (inspiration), and carbon dioxide, a waste product, leaves the body during breathing out (expiration).

White blood cells normally found in our bodies help protect us from infection. But white blood cells also release an enzyme,

called neutrophil elastase, that can damage the lungs. In normal lungs, alpha-1 antitrypsin protects the lungs from the harmful effects of neutrophil elastase.



What happens if there isn't enough alpha-1 antitrypsin?

When the lungs do not have enough alpha-1 antitrypsin, neutrophil elastase is free to destroy lung tissue. As a result, the lungs lose some of their ability to expand and contract (elasticity). This leads to emphysema and sometimes makes breathing difficult. Shortness of breath may occur.

The speed at which lung tissue is destroyed varies with each person. What is known, though, is that tobacco smoking worsens the lung damage.

How does smoking worsen lung damage caused by the disorder?

Tobacco smoke irritates and damages the lungs, prompting the body to send more white blood cells to protect them. The more white blood cells there are, the more neutrophil elastase is made, causing even more lung damage.

Also, the smoke itself changes alpha-1 antitrypsin so that it cannot do as good a job protecting the lungs from harm.

Smokers with alpha-1 antitrypsin deficiency have a faster rate of lung damage. So if you smoke, *stop*.

What are the risk factors for the disorder?

Alpha-1 antitrypsin deficiency is not contagious, and you cannot “catch it” from someone. The disorder is inherited, which means that it is passed on genetically from a relative. All persons who have relatives with this disorder should consider being tested to find out whether they carry the gene for it.

How is the disorder inherited?

Everyone receives one gene for alpha-1 antitrypsin from each parent. The M gene is the most common type of gene, and it is normal. The person who inherits an M gene from each parent has normal levels of alpha-1 antitrypsin.

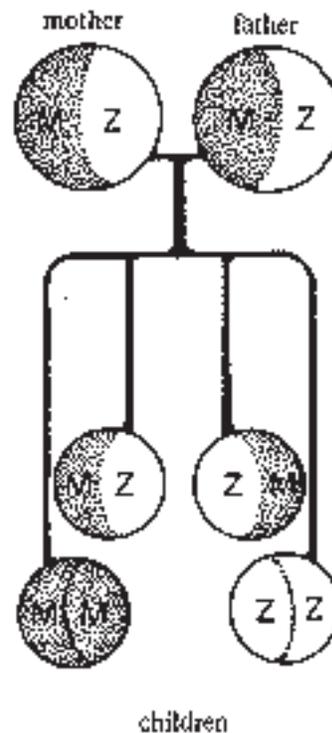
The Z gene is the most common defect that causes the disorder. If a person inherits one M gene and one Z gene, that person is a carrier of the disorder. While such a person may

not have normal levels of alpha-1 antitrypsin, there should be enough to protect the lungs.

The person who inherits the Z gene from each parent is called “type ZZ.” This person has very low alpha-1 antitrypsin levels, which allow neutrophil elastase to damage the lungs.

In rare cases, a person’s body may not produce *any* alpha-1 anti-trypsin. This condition is also inherited, and it is called “null-null type.”

Another type, called “dysfunctional,” means that although alpha-1 antitrypsin levels are normal, alpha-1 antitrypsin does not work the way it should. This type of the disorder is very uncommon.



This diagram shows the most common way the disorder is inherited. There are four possible combinations of passing the Z and M genes from parents to children.

What are the signs and symptoms of alpha-1 antitrypsin deficiency?

A person with this disorder can be short of breath during daily activities. This is because the air sacs have been destroyed, and the lungs trap air as they expand and contract during breathing.

Can the disorder be treated?

There are several ways you can protect your lungs from the effects of the disorder:

- receive immunizations for flu and pneumonia
- receive early treatment for lung infections by seeing your doctor at the first sign of a cold or other lung problem
- avoid tobacco smoke, noxious fumes, dust, and pollution
- stay fit by doing regular exercise
- increase your alpha-1 anti-trypsin level.
Speak with your doctor about alpha-1 antitrypsin replacement therapy.

You can also reduce symptoms of shortness of breath by doing the following:

- using medications (for example, bronchodilators or inhaled steroids) prescribed by your doctor to help open your airways
- using oxygen if your doctor prescribes it
- doing pulmonary rehabilitation (including breathing techniques). Call your local lung association to find out more.

Research is being conducted at NIH to learn more about alpha-1 antitrypsin deficiency. Scientists are studying how the disorder affects the body as well as new and future treatments. Someday, gene therapy may be able to fix the inherited problem.

If you have questions about alpha-1 antitrypsin deficiency, feel free to ask your nurse or doctor.



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