
Understanding Alpha-1 Antitrypsin Deficiency (A1AD)

Alpha-1 antitrypsin deficiency (A1AD) is a rare genetic disorder. People who have A1AD have low levels of alpha-1 antitrypsin (A1A) in their blood. A1A is a protein. This protein is normally made in the liver. Not having enough of the A1A protein lets an enzyme called elastase build up. The extra enzyme then attacks other tissues in the body. It may attack tissue in the lungs, liver, and skin. It can lead to a lung disease called emphysema. This is a type of chronic obstructive pulmonary disease (COPD). It can also lead to liver disease (cirrhosis). But some people with A1AD don't develop these problems.

What causes A1AD?

A1AD is caused by a change (mutation) in a gene that is passed on in families. This means a person is born with it. A1AD is rare. It happens more in people whose family is from Europe.

Symptoms of A1AD

A1AD doesn't cause symptoms unless tissue is damaged.

People with A1AD may develop emphysema. This is a lung disease where the small air sacs in the lungs (alveoli) become damaged. Smoking raises the risk of emphysema. Symptoms of emphysema may start between ages 20 and 50. They can include:

- Trouble catching your breath with mild activity
- Wheezing
- Tiredness
- Weight loss
- A chronic cough with phlegm
- Chest colds or infections (pneumonia) that happen often

A1AD can cause liver disease. It can lead to scar tissue in the liver (cirrhosis). This causes symptoms such as:

- Tiredness
- Loss of appetite
- Weight loss
- Yellow color in skin or eyes (jaundice)
- Swollen belly
- Swollen legs
- Blood in vomit or stool

In some people, A1AD may lead to inflammation of fat under the skin. This is called panniculitis. It causes lumps under the skin that are red and painful.

Diagnosing A1AD

To diagnose A1AD, your healthcare provider will give you an exam and ask about your health history. You will have a blood test. This is to check for the gene change. You may have a chest X-ray or other imaging test to look for other problems. Your lungs may be tested to check how well they work. If you are diagnosed with A1AD, your family members should have the blood test to check for the gene change. Only about 1 in 10 people with A1AD have been diagnosed. Family members with A1AD can make lifestyle changes to help prevent serious problems.

Treatment for A1AD

You and your healthcare provider can discuss the best plan to treat your condition.

Treatment for emphysema may be done with any of these:

- Inhaled bronchodilator medicine
- Inhaled steroid medicine
- Anticholinergic medicine
- Oxygen therapy
- Antibiotics or other medicines for lung infections

Some people with emphysema may be able to do augmentation therapy. This is done through an IV into a vein. Blood plasma with a lot of A1A protein is sent into the blood. This may be done once a week.

If your lung disease or liver disease is severe, your healthcare provider may advise a lung or liver transplant. A transplant is surgery to remove a diseased organ and replace it with a healthier one. Your provider will talk with you about this type of surgery if needed.

Possible complications of A1AD

People with A1AD are at risk of hepatocellular carcinoma. This is a type of liver cancer. You may need tests over time to check for this. You may have a biopsy. This is done by taking a small sample of liver tissue. The tissue is examined for cancer cells.

Living with A1AD

Not every person with A1AD will develop emphysema. Here are some ways you can help prevent problems:

- Don't smoke.
- Don't be around people who smoke.
- Don't live or work in places with a lot of dust or chemicals.
- Don't use aerosol sprays.

You are more likely to develop emphysema if you have any of these:

- Asthma
- A lot of lung infections
- A family history of emphysema

If you have emphysema, make sure to:

- **Prevent lung infections.** You can do this with a yearly flu vaccine. You should also get a pneumonia vaccine as often as advised.
- **Keep a healthy lifestyle.** Get regular exercise that helps your lungs. Eat healthy foods to give your body good nutrition.

You may need tests over time to check the health of your liver. You may have blood tests. You may have imaging tests such as ultrasound. Your healthcare provider will tell you how often to have these.

If you have liver disease:

- Don't drink any alcohol.
- Ask your healthcare provider what medicines are safe to take. Some can harm the liver. This includes acetaminophen.
- Talk with your provider before taking any vitamins, herbs, or other supplements.

When to call your healthcare provider

Call your healthcare provider right away if you have any of these:

- Symptoms that get worse
- New symptoms