Alpha-1 antitrypsin deficiency

Alpha-1 antitrypsin (AAT) is a protein made mainly by your liver. It protects your lungs and other organs from infections and irritants such as cigarette smoke and gasses.

Alpha-1 antitrypsin deficiency is where people have low levels of AAT in their bloodstream. It is a rare genetic (inherited) disorder. The disorder can increase your risk of developing lung and liver diseases, including emphysema (damaged air sacs in the lungs) and cirrhosis (scarring of the liver).

AAT deficiency can cause lung problems in adults and liver problems in both adults and children. Some of these conditions can be life-threatening.

In Ireland, 1 in 25 people carries the gene that causes AAT deficiency.

Symptoms of AAT deficiency

People with AAT deficiency may not develop related diseases and may have no symptoms. You might never know you have the disorder. People who have symptoms usually notice them between ages 20 and 50.

The symptoms of AAT deficiency are like those caused by chronic obstructive pulmonary disease (COPD).

Symptoms may include:

- increasing shortness of breath
- a chesty cough with phlegm that never seems to go away
- frequent chest infections
- ongoing wheezing

Symptoms may increase when you have a cold or flu.

When the liver is affected by AAT deficiency, symptoms may include:

- tiredness
- loss of appetite
- weight loss
- swelling of the feet or belly
- yellowish discolouration of the skin (jaundice) or the white part of the eyes
- vomiting of blood

- blood in your poo

Talk to your GP if you have these symptoms.

Diagnosing AAT deficiency

AAT deficiency runs in families. Many people do not know that they have it. Early diagnosis can help prevent serious lung disease.

Treatment for AAT deficiency

AAT deficiency has no cure. You may need oxygen therapy, pulmonary rehabilitation, or medicines to treat complications. If you smoke, you should stop. You should also avoid secondhand smoke and other harmful fumes.

If you have complications from AAT deficiency, you may also need:

- medicines called inhaled bronchodilators and inhaled steroids - these medicines help open your airways and make breathing easier
- oxygen therapy
- pulmonary rehabilitation
- a lung transplant - only if you have very severe breathing problems and have a good chance of surviving the transplant surgery

Living with AAT deficiency

If you have AAT deficiency, you may have a related lung or liver disease. Ongoing medical care and lifestyle changes can help you manage your health.

Regular follow-up care is important.

You will need to:

- have regular pulmonary (lung) function tests or imaging tests to monitor your condition
- get the flu and pneumococcal vaccines to protect you from illnesses that may worsen your condition
- continue to take all your medicines as prescribed and follow your treatment plan

If you have a lung infection or any breathing problems you will need immediate treatment.

The Alpha-1 foundation Ireland website has more information and supports for living with AAT.

Page last reviewed: 21 September 2021
Next review due: 21 September 2024

HSE Live - we're here to help

Monday to Friday: 8am to 8pm
Saturday: 9am to 5pm
Sunday: Closed
Bank holidays: Closed

Freephone: 1800 700 700

From outside Ireland: 00 353 1 240 8787

HSE Facebook

HSE Instagram

HSE TikTok

HSE YouTube

HSE LinkedIn

HSE X (Twitter)

Complaints and feedback

Emergencies

Cookie settings