

# What is alpha-1-antitrypsin deficiency?

FACT SHEET prepared by The Asthma and Respiratory Foundation NZ

**Alpha-1-antitrypsin deficiency is an inherited disorder, which usually affects the lungs. With a severe deficiency liver damage may also occur.**

## What is alpha-1-antitrypsin?

Alpha-1-antitrypsin is a protein, which is produced by the liver and enters the blood stream. Its main role is to protect the lungs from damage caused by enzymes. These enzymes are part of the lungs defences against infection, but if these are not balanced by alpha-1-antitrypsin, damage to the lung tissue can occur.

## What is alpha-1-antitrypsin deficiency?

Alpha-1-antitrypsin deficiency becomes important when the concentration of the protein in the blood is less than 20% to 30% of what we would normally expect. When this occurs, the lung is poorly protected from destructive enzymes and loss of lung tissue occurs, leading to a condition called emphysema. It is estimated that one in every 2,500 people have severe alpha-1-antitrypsin deficiency (levels below 20% of normal). This means that approximately 1,600 New Zealanders have alpha-1-antitrypsin deficiency.

## What is emphysema?

In emphysema the air sacs (alveoli) in the lungs are damaged and eventually destroyed. This leads to difficulty in absorbing enough oxygen. The main symptom of emphysema is breathlessness, which gets worse as the emphysema progresses. Emphysema is irreversible — it cannot be 'cured', but the symptoms can be controlled.

The major cause of emphysema is cigarette smoking. People with an inherited alpha-1-antitrypsin deficiency who smoke are likely to develop emphysema at a young age and the disease will progress at a much faster rate.

## Can I have an alpha-1-antitrypsin deficiency and not get emphysema?

Yes, everyone has different amounts of alpha-1-antitrypsin protein in their blood. Not all people with low levels develop emphysema, as the amount of alpha-1-antitrypsin present will still be enough to protect the lung from destruction in most cases. Emphysema is much more likely to occur if you smoke. Most, but not all people with levels less than 30% of normal will develop emphysema. Levels above 30% of normal seem to give adequate protection unless the person smokes.

## Can I be treated for alpha-1-antitrypsin?

There is no specific treatment for alpha-1-antitrypsin deficiency at present. Some people find that inhaled reliever medication (Atrovent, Bricanyl, Respigen, Salamol or Ventolin) helps relieve their symptoms. You will need to discuss this with your doctor. It is also very important to avoid prolonged exposure to dust, fumes and especially cigarette smoke. These pollutants can accelerate the rate at which lung tissue is destroyed.

### **Research and development of new treatment**

Replacement treatment with intravenous infusions of alpha-1-antitrypsin concentrate is now possible and sometimes used overseas. Higher blood concentrations of alpha-1-antitrypsin would hopefully protect lung tissue from destruction. This treatment is still very much in the experimental stage as the treatment has not yet been proven to have long-term benefits. At present this treatment is not available in New Zealand.

### **The Asthma and Respiratory Foundation NZ can help you**

The Asthma and Respiratory Foundation NZ is New Zealand's not-for-profit sector authority on asthma and other respiratory illnesses. We advocate to government and raise awareness of respiratory illnesses, fund research for better treatments and educate on best practice.

Check out our comprehensive website at [asthmaandrespiratory.org.nz](http://asthmaandrespiratory.org.nz)

For further information on alpha-1-antitrypsin deficiency visit [lungnet.org.au](http://lungnet.org.au) and [lunguk.org](http://lunguk.org)

The Asthma and Respiratory Foundation NZ would like to acknowledge the Australian Lung Foundation for the information contained in this fact sheet.

### **References**

The Australian Lung Foundation

British Lung Foundation, London