

# Fact Sheet - Lung Disease & Alpha-1 Antitrypsin Deficiency

## Background

Alpha-1 antitrypsin deficiency (A1AD) is a common serious genetic disorder, passed on from parents to their children. A1AD is found in most countries but usually in people with a European background (e.g. Scandinavian, British, Spanish). A1AD occurs in one in nine people in Australia and can cause serious lung and liver disease. However, many doctors are poorly informed about A1AD. The Alpha-1 Organisation Australia (A1OA) is working to change this situation.

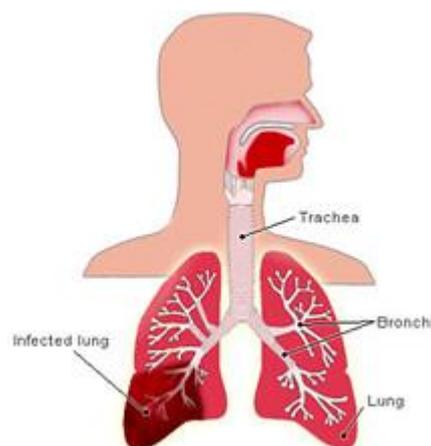
## The Role of the Lungs

Lungs bring oxygen into your body and remove carbon dioxide and other waste gases through gas exchange. Gas exchange occurs in the capillaries surrounding the air sacs (the alveoli) which are at the end of bronchioles in your lung. Bronchioles are connected to larger air tubes (bronchi). The bronchi join onto the trachea (wind pipe). Every cell in your body needs oxygen. If your respiratory system doesn't work properly you can feel breathless, fatigued and organs can be damaged. Lung disease means that parts of the lungs are inflamed or damaged and aren't working as well as they should.

## What Causes Lung Disease in A1AD?

A1AD lung disease is found in some adults (not children). About 50% of alphas with a severe AAT deficiency will have lung disease. Alphas are commonly misdiagnosed with adult asthma. Symptoms commonly present between the ages of 20 – 50 but can occur later.

The reason why not all alphas develop lung disease is not understood. Alpha-1 antitrypsin (AAT) is a protein made mostly by the liver. It is



**Pneumonia in the right lung**

Image source: "[pneumonia w/ pleurisy, right lung](#)" by [coreythrace](#) is licensed under [CC BY-NC 2.0](#)

meant to leave the liver and circulate around the body to protect other organs, but, if the AAT is stuck in your liver, or you don't make enough, your lungs can be attacked by a natural protein called neutrophil elastase which is released from white blood cells to fight infection such as pneumonia. Currently there is no way to help the protein leave the liver but clinical trials are underway. Normally, neutrophil elastase plays an important role in clearing out bacteria and keeping lungs healthy. If you don't have enough AAT the neutrophil elastase is not kept in balance, then lung disease can occur e.g. emphysema and loss of lung elastin. Elastin provides the elasticity to the structure of the lung. Lung disease can make it harder to clear sputum from the lung causing more problems.

## Lung Hyperinflation

The lungs hyperinflate when air gets trapped in them and causes them to overinflate. Hyperinflation can occur when air sacs are damaged and are less elastic, making it difficult to breath out all of the air from the lungs.

## Emphysema

Without enough AAT, neutrophil elastase can destroy alveoli and cause emphysema prematurely. Emphysema is the permanent expansion of the airway. The Z gene is most commonly associated with emphysema in alphas. Emphysema can be made worse by smoking, exposure to gas, dust and chemicals, so alphas are advised to avoid these triggers.

## Bronchiectasis

Another lung disease called bronchiectasis is associated with A1AD. This disease is associated with inflammation in the airways, scarring, increased mucus, and increased neutrophil activity.

## Chronic Bronchitis

Chronic bronchitis is associated with A1AD. It is chronic inflammation of the airways that causes increased sputum production. Patients with A1AD have greater inflammation. It is thought that the airways are not protected from neutrophils which causes more inflammation.

## Non-Tuberculosis Mycobacterium

People with lung disease such as chronic bronchitis and bronchiectasis are at higher risk of having a bacteria called non-tuberculosis mycobacterium (NTM) in their lungs. Your doctor should recommend testing your mucus for NTM and will advise if treatment is needed.

## Symptoms of Lung Disease

- Shortness of breath (dyspnea)
- Excessive cough with phlegm
- Wheezing
- Frequent chest infections
- Fatigue
- Decreasing in exercise capacity
- Coughing up blood
- Asthma like symptoms
- Low grade fever
- Night sweats
- Chest pain (worse when breathing in)

## Treatment for Lung Disease

The American Thoracic Society suggests weekly infusion for everyone with FEV1 less than 80% predicted. Unfortunately, augmentation therapy

(a weekly infusion of plasma containing antitrypsin) is not subsidised by the Australian Government but can slow or stop the progression of emphysema. To purchase augmentation therapy privately in Australia, your doctor will have to apply to the Therapeutic Goods Administration (TGA). The cost is based on body weight – approx. \$100,000 per annum.

Other treatments include:

- Rapid antibiotic treatment for lung infections to help stop the damage done by neutrophil elastase
- Lung valves to block off damaged areas of the lung (e.g. Zephyr valves) to help breathing
- Double lung transplant

## Activities to Support Lung Health

The most important activity is to avoid cigarette smoking. Other things that you can do:

- Attend pulmonary rehab
- Exercise
- Practice lung health activities (see our Fact Sheet Lung Health)
- Treat lung infections immediately
- Avoid sick people (lung disease is associated with lung infections)

## What Tests Can Find Lung Disease?

- X-ray
- Chest CT scan
- Pulmonary function tests (e.g. spirometry, FEV1, DLCO)

## Need More Information or Support?

Contact Alpha-1 Organisation Australia : email [contactus.a1oa@gmail.com](mailto:contactus.a1oa@gmail.com)

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This brochure is one in a series of information sheets produced by the Alpha-1 Organisation Australia (A1OA). This information is designed to be a guide only and does not replace advice given by your health professional. Any treatment information or brand names are correct at the time of printing. If the information raises concerns or if you have further questions please consult your doctor.