

# Alpha1-antitrypsin deficiency



Lung  
Foundation  
Australia

## Overview

Alpha-1 antitrypsin (AAT) is a protein made in the liver and released into the bloodstream where it moves into the lungs. It helps protect the lungs from damage caused by infection and inhaled irritants, such as tobacco smoke.

Alpha1-antitrypsin deficiency (AATD) is a condition where the liver makes too little AAT. As a result, the level in the lungs is too low to protect against damage, potentially leading to the development of lung disease such as Chronic Obstructive Pulmonary Disease (COPD). More rarely, it can cause liver or skin disease.

## Cause

AATD is an inherited genetic condition. Everyone has two copies of the gene responsible for AAT protein production in the liver. Each parent passes one of the two possible AAT genes to their child. If you receive one faulty gene from either of your parents, you are considered to be a carrier of a defective AAT gene. Your body should still produce enough AAT to protect your lungs, however, some people with one faulty gene may develop lung problems if they smoke.

If both parents pass on a faulty gene, you will almost certainly develop AATD. If you have AATD you are more likely to develop COPD. This is likely to be diagnosed at a younger age than usual and it may progress more rapidly than COPD that is not attributed to AATD. Smoking is the most crucial risk factor in contributing to the development of COPD for people with AATD. Exposure to secondhand smoke, working in a dusty environment, having asthma or a history of repeated lung infections can also increase the risk of developing COPD.

## How common is AATD?

AATD is a disease that can affect males and females of all ages and ethnic backgrounds. It is estimated there are 30,000 people in Australia and New Zealand with AATD, however, less than 10% of these people have been diagnosed<sup>1</sup>. Around 1% of all patients with COPD have AATD.

## Symptoms

### Common symptoms of AATD-related COPD include:

- Shortness of breath (breathlessness)
- A repetitive cough that doesn't get better
- Increased phlegm or mucus production
- Feeling tired or low energy
- Low exercise tolerance
- More frequent chest infections or taking longer to recover from a cold or chest infection.

## Diagnosis

AATD is diagnosed via a simple blood test which measures the level of AAT in your blood. Your doctor will likely also perform a physical examination and ask about your family history. The symptoms of AATD are often associated with other conditions which can lead to delays in diagnosis. If you have early onset COPD, or you have parents or close relatives who have been diagnosed with AATD, it is worthwhile considering getting tested for the condition.

Although there is no current cure for AATD, early diagnosis and treatment of AATD related COPD is important as it enables you to make positive lifestyle changes to protect your lung health, which can help to slow the progression of the disease and improve how you feel. You may also be prescribed inhalers to treat your symptoms and help to reduce the risk of flare-ups.

## Treatment

Currently, augmentation therapy is the only treatment specifically designed for AATD. It has been approved for use in Australia, however the cost is not covered by Medicare. This treatment involves intravenous infusions of AAT concentrate to correct the deficiency in the blood and lungs. Evidence about how this treatment impacts outcomes such as symptom severity and breathing test results is still emerging. Speak with your respiratory specialist about the best options for you and your specific symptoms.

## Self-management

**If you have been diagnosed with an AATD-related COPD, there are many actions you can take to stay as well as possible:**

### Quit smoking

Research shows that people with AATD who smoke experience a greater decline in lung function than ex-smokers or those who have never smoked. Not all people with AATD-related COPD will have smoked, but if you do, quitting is the single most important thing you can do to improve your health and lung function.

### Keep your vaccinations up to date

Discuss with your doctor the seasonal influenza, pneumococcal pneumonia and COVID-19 vaccination.

### Have an up-to-date COPD Action Plan

Develop a written plan with your doctor to help you understand your day-to-day symptoms, how to recognise when your symptoms change and what action you need to take.

## Be physically active

Regular exercise can help maintain your fitness and wellbeing as well as improve your symptoms and quality of life. You should aim for at least 30 minutes, five times a week. Before starting any new exercise program, talk to your doctor who can refer you to a physiotherapist or exercise physiologist to provide advice on a program that is right for you.

### - Pulmonary rehabilitation

Pulmonary rehabilitation is an exercise and education program provided by specially trained health professionals. It teaches you the skills you need to exercise safely, manage your breathlessness and to stay well and out of hospital.

### - Lungs in Action

Lung Foundation Australia's Lungs in Action program is a safe and fun community-based exercise class designed to help people with a chronic lung disease maintain the benefits achieved through pulmonary rehabilitation.

## Eat well

Diet greatly impacts your general health and maintaining a healthy weight range is important. You should enjoy foods from a wide variety of nutrition sources. If you need support with maintaining a healthy diet, ask your doctor for a referral to a dietitian.

## Access emotional support

Connect with friends, family and stay engaged with activities where possible. Anxiety and depression are common for people living with a lung disease. It is important to access support to help maintain your mental health and wellbeing. Talk to your doctor or contact Lung Foundation Australia's Information and Support Centre for more information.



## FURTHER INFORMATION AND SUPPORT

Contact Lung Foundation Australia for more information, to access our support services and join our mailing list for regular updates and latest news.

### Lung Foundation Australia Services

- Information and Support Team
- Lung disease information resources
- Education webinars
- Lung Cancer and Respiratory Support Nurses
- Support groups
- Peer-to-peer connections
- Referral to pulmonary rehabilitation and Lungs in Action exercise programs
- E-newsletter

### Alpha-1 Support Links

- Alpha-1 Association of Australia  
[www.alpha1.org.au](http://www.alpha1.org.au)
- Alpha-1 Organisation Australia  
<https://a1oa.org.au/>

[Lungfoundation.com.au](http://Lungfoundation.com.au) | Freecall 1800 654 301 | [enquiries@lungfoundation.com.au](mailto:enquiries@lungfoundation.com.au)

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**References:** 1 Dummer J, Dobler CC, Holmes M, et al. Diagnosis and treatment of lung disease associated with alpha one-antitrypsin deficiency: A position statement from the Thoracic Society of Australia and New Zealand. *Respirology*. 2020;25(3):321-335. doi:10.1111/resp.13774 Viewed 25 August 2020.