

Past and present supporters of the Alpha-1 Association of Australia include the following organisations:



ALPHA-1 Association of Australia

Alpha-1-Antitrypsin Deficiency

Do you have any of these or related symptoms?

Lungs

Early onset emphysema

Chronic bronchitis

Recurrent chest colds

Liver

Jaundice

Unexplained abnormal liver function

WHAT IS ALPHA-1-ANTITRYPSIN DEFICIENCY (A1AD)?

Alpha-1-Antitrypsin Deficiency is a genetic disorder which predisposes an individual to several illnesses which affect mainly the lungs and liver. Such diseases include emphysema, liver cirrhosis, or more rarely, panniculitis (a skin condition) or vasculitis (inflammation of blood vessels).

A1AD is caused by the deficiency of a protein called alpha-1 antitrypsin (AAT) in the bloodstream. AAT is produced by the liver and protects delicate lung tissue from an enzyme called neutrophil elastase (NE). The normal behaviour of NE is to remove damaged cells, but when its activity levels become too high it will also attack healthy lung tissue.

In A1AD, AAT is retained in the liver, leading to a shortage and loss of protection from NE in the lungs. Infections and exposure to smoking can also exacerbate lung damage. At the same time, the AAT accumulated in the liver can also damage liver tissue.

WHO IS AFFECTED BY A1AD?

As a genetic condition, A1AD is passed from parents to children through genes. Everyone has two copies of the AAT gene, most people have two normal copies. A1AD sufferers may have either one normal and one disease-associated copy, or two disease-associated copies of the gene.

Approximately 1 in 50 people worldwide have at least one affected gene*. The severity of the disease varies between individuals, as AAT levels in the bloodstream may range between 10-80% of normal levels, depending on the number and type of affected genes an individual has.

Please note: The information in this brochure is intended as a guide only and is not an authoritative statement. Please consult your doctor if you have further questions.

*Estimate based on data from: de Serres, (2002), Chest, v.122, p1818-1829

WHAT ARE THE SYMPTOMS OF A1AD?

A1AD affects people in different ways and symptoms can vary between individuals. Some common early symptoms, associated with lung or liver dysfunction, include:

- excess sputum (phlegm)
- shortness of breath
- coughing and/or wheezing
- recurrent chest colds or lung infections
- history of suspected allergies
- asthma
- feeling fatigued
- decreased appetite
- abdominal swelling
- abnormal liver function tests
- enlarged liver or spleen
- skin lesions or inflammation

CAN A1AD BE TREATED?

Currently there is no specific treatment for A1AD in Australia aside from standard procedures to relieve the symptoms of the associated illnesses. However, with early diagnosis and by making particular lifestyle adjustments, affected individuals can minimise the effects of A1AD. Such measures include:

- avoiding tobacco smoke, noxious fumes, dust and pollution
- receiving early treatment for lung infections and colds
- receiving immunisations against influenza and pneumonia
- staying fit with regular exercise
- moderating alcohol consumption
- maintaining a healthy and balanced diet, ensuring the liver is supplied with essential nutrients

MEDICAL TESTS AND DIAGNOSIS OF A1AD

A1AD is one of the most common hereditary disorders in the world, but it often remains undiagnosed. If you suffer from any symptoms listed in this brochure you can ask your doctor for a simple and inexpensive test to measure the level of AAT in your bloodstream. Low AAT levels may indicate a need for DNA testing to confirm A1AD and an individual's AAT genetic make-up.

If you have a relative who has been diagnosed with A1AD or a family history of early emphysema or liver disease, DNA testing is also recommended, even if your AAT levels are not especially low.

Some people with A1AD may present without obvious symptoms and lead relatively healthy lives, especially if they avoid smoking. However, they still risk passing the affected gene on to their children. These people should also seek medical attention should early symptoms of A1AD become apparent.

For more information about A1AD and support:

Alpha-1 Association of Australia

Web site and online discussion group:

<http://www.alpha1.org.au>

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