



ALPHA-1 Association of Australia

## **NON SMOKERS WITH AIRWAYS DISEASE OR ASTHMA SUFFERERS UNRESPONSIVE TO TREATMENT?**

**Are you treating patients with airways disease who have not smoked?  
Is your asthmatic patient not responding to treatment?  
Could they be suffering from Alpha-1 Antitrypsin Deficiency?**

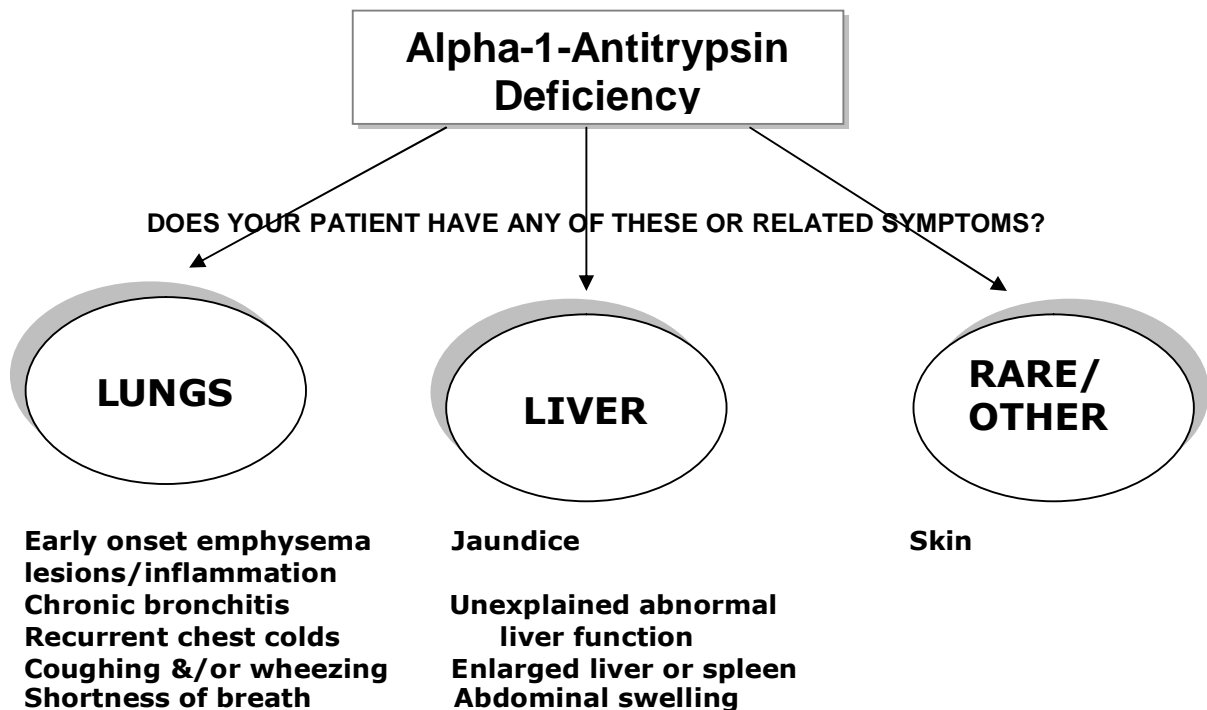
Alpha-1 antitrypsin deficiency (A1AD) is a rare cause of emphysema. It is a genetic condition in which the serum concentrations of the protein alpha-1 antitrypsin (A1AT) are reduced or very occasionally completely absent. A1AT acts as an antiprotease and helps to protect the lungs from other harmful enzymes (proteases) that destroy lung tissue. In normal circumstances proteases and antiproteases exist in a harmonious balance. However, if this balance is upset in favour of the proteases then tissue damage occurs and in the lung emphysema develops.

A1AD may also cause with other illnesses which can affect the liver, such as liver cirrhosis, or more rarely, panniculitis (a skin condition involving inflammation of subcutaneous tissues).

### **THE SYMPTOMS OF A1AD**

A1AD affects people in different ways and symptoms can vary between individuals. Some of the common early symptoms, associated with lung or liver dysfunction, are shown in the Figure.

*With some symptoms similar to asthma it is not unusual for A1AD not to be detected and for the patient to be treated for asthma in error.*



### **WHO IS AFFECTED BY A1AD?**

There are many different types of A1AT. The various different types are called phenotypes and are determined by genetic messages. Very few of these phenotypes lead to a blood A1AT concentration low enough to cause disease. The commonest of these is known as PiZZ.

### **WHO SHOULD BE TESTED FOR A1AD**

A1AD is one of the most common hereditary disorders in the world, but it often remains undiagnosed. Patients who suffer from any symptoms listed above or who are not responding to treatment for unexplained chronic airways disease or asthma should have a simple and inexpensive test to measure the level of A1AT in their bloodstream. Low blood A1AT levels should be followed by phenotype testing. (*See Medicare Benefits Schedule – Item 66635 – Quantitation in serum, urine or other body fluid and Item 66638 – Isoelectric focussing or similar methods for determination of Alpha-1-antitrypsin phenotype in serum.*)

Testing is recommended for relatives of persons with diagnosed A1AD or a family history of early emphysema or liver disease. Note that ***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. Such persons should be given medical advice about how to minimise their risk of developing significant health problems, eg avoid smoking to minimise the chance of developing emphysema.

### **CAN A1AD BE TREATED?**

Currently there is no specific treatment for A1AD in Australia. Treatment is aimed at symptom relief and reducing the chance of progressive disease.

Such measures include:

- not smoking
- avoiding tobacco smoke, noxious fumes, dust and pollution
- receiving early treatment for respiratory tract infections
- 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.

### **WHAT HELP IS AVAILABLE?**

For those who have been diagnosed and/or their family and carers, help is available via the Alpha-1 Association of Australia (AAA). The Association provides information on the illness which is updated from time to time. An online discussion group enables members to exchange information, tips and personal advice which can be of particular benefit to the newly diagnosed.

#### **Alpha-1 Association of Australia**

Web site and online discussion group:

[www.alpha1.org.au](http://www.alpha1.org.au)

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As with other forms of Chronic Obstructive Pulmonary Disease, it is important to identify A1AD as soon as possible so that the condition can be managed early with consequently better outcomes.