

WHO IS AFFECTED BY A1AD?



A1AD genes are passed from parents to children. Everyone has two copies of the AAT gene. Most people have two normal copies. If you have a normal gene and one abnormal gene, there may be some deficiency, but A1AD sufferers usually have two disease-associated copies of that gene.

The genes are the coded plans for the cell to make AAT, so if the genes are faulty, the amount of AAT made is low or faulty. The severity of any problem depends on how much AAT is made and released into the bloodstream. The less that is present in the blood, the more trouble results. AAT levels in the bloodstream may range between 10-80% of normal levels. For emphysema to result, the level is usually well down. In NZ, the laboratory normal range is quoted as 1.2-2.0 gm/litre. Problems are not usually seen until levels are under 0.8, and more usually as low as 0.5. Levels of 0.3 or less are often seen in individuals with severe A1AD. The levels depend on the kind of abnormal gene the individual has.

Remember, though, that not everyone even with low levels of AAT will get emphysema, as the additional factor of airway inflammation (from smoke or dust or other conditions) is needed to cause damage.

For more information and support

EMAIL alpha1.info@nzord.org.nz

Dr Val Orchard
Wellington

E valjim@actrix.co.nz

P +64 4 5287050

M 0220631167

Marilyn Black
Manawatu

E mandblack@gmail.com

P +64 6 3289018

M 0273442999

Other Support

The NZ Asthma Foundation
W asthmafoundation.org.nz

Alpha-1 Foundation USA
W alpha-1foundation.org

Alpha-1 Association Australia
W alpha1.org.au

Peer review by
Doctor David Jones - Respiratory Specialist Wellington

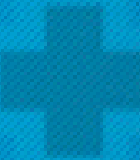
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NZORD -The NZ Organisation for Rare Disorders | nzord.org.nz

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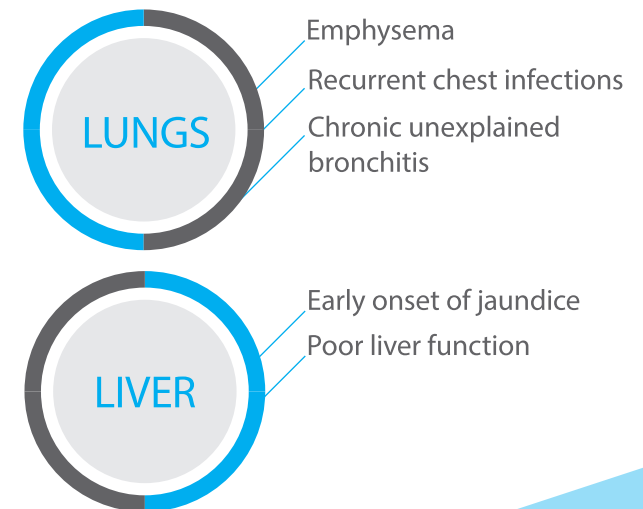
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Alpha-1 Antitrypsin Deficiency (A1AD)



DO YOU HAVE ANY OF THESE SYMPTOMS?



WHAT IS ALPHA-1 ANTITRYPSIN DEFICIENCY?

Alpha-1 antitrypsin deficiency (A1AD) is a genetic disorder that can produce emphysema in the lungs, cirrhosis of the liver, and occasionally inflammation in the fat layer of the skin (called panniculitis).

Alpha-1-antitrypsin (AAT) is a protein made in the liver that then circulates in the blood. Its job is to protect the lungs from the inflammation produced when the body's immune system fights infection and inhaled irritants such as tobacco smoke. This battle by the immune system against inhaled particles and germs will result in release of digestive enzymes from the broken down cells. These enzymes will damage the lung tissue itself unless they are mopped up and disposed of, which is exactly the job that AAT does. Without it, lung damage results, in the form of emphysema.

The shortage of AAT in the blood happens when the AAT protein made in the liver is abnormal and does not get released by the liver cells at the normal rate. The build up of AAT in the liver cells causes damage leading to liver disease.

AAT Genetic testing is recommended if you have a relative who has been diagnosed with A1AD or a family history of early emphysema or liver disease.

WHAT ARE THE SYMPTOMS OF A1AD

A1AD affects people in different ways and symptoms can vary between individuals.

WITH EMPHYSEMA

Shortness of breath is the main symptom, slowly progressing to become very limiting. Also, Cough and phlegm and repeated chest infections, and possibly wheezing. Standard asthma inhalers tend not to be as useful as hoped.

WITH LIVER DAMAGE

Liver damage leading to cirrhosis can cause:

- Abnormal liver tests on blood testing.
- Nausea
- Jaundice (yellow skin and eyes)
- Swelling of the abdomen and large liver and spleen.

CAN A1AD BE TREATED?

The treatments offered at present are the same as for people who have emphysema for other reasons. Inhalers and other medications can help. However, it is important to address the following lifestyle measures:

- **Do not smoke.**
- **Keep physically active with regular exercise**
- **Keep body weight about right (avoiding getting big but also avoiding losing muscle bulk)**
- **Have Flu vaccination and pneumococcal vaccination**
- **Get any chest infections treated promptly**
- **Avoid situations that irritate the chest and that can make emphysema worse, ie smoke, fumes, dust,pollution.**

Currently there is no actual cure for A1AD. In some countries (not in NZ), AAT made from donated blood is administered regularly by intravenous infusion - this is called augmentation therapy. The benefits/downsides of this therapy are being evaluated currently by an international medical research trial in New Zealand and Australia.

Some people with advanced emphysema are eligible for lung transplantation. However with early diagnosis and lifestyle adjustments, affected individuals can reduce the likelihood of reaching that level of severity.

MEDICAL TESTS AND DIAGNOSIS OF A1AD

A1AD often remains undiagnosed. Lung symptoms may not show until people are in middle age and can be misdiagnosed as asthma. If you begin to suffer from the symptoms listed in this brochure you can ask your doctor for a simple and inexpensive blood test to measure the level of AAT in your blood. Low AAT levels will lead to testing the individual's AAT genetic make-up. Referral then to see a respiratory specialist is usual so that regular x-rays and lung function tests can monitor you and medications applied to help symptoms.

AAT Genetic testing is also recommended if you have a relative who has been diagnosed with A1AD or a family history of early emphysema or liver disease. Early warning for a young person can guide them to avoid smoking and to choose the right sort of work.

Some people with A1AD may not have obvious symptoms and can be leading healthy lives, especially if they avoid smoking. However, they still risk passing an affected gene onto their children and can develop symptoms in later life.