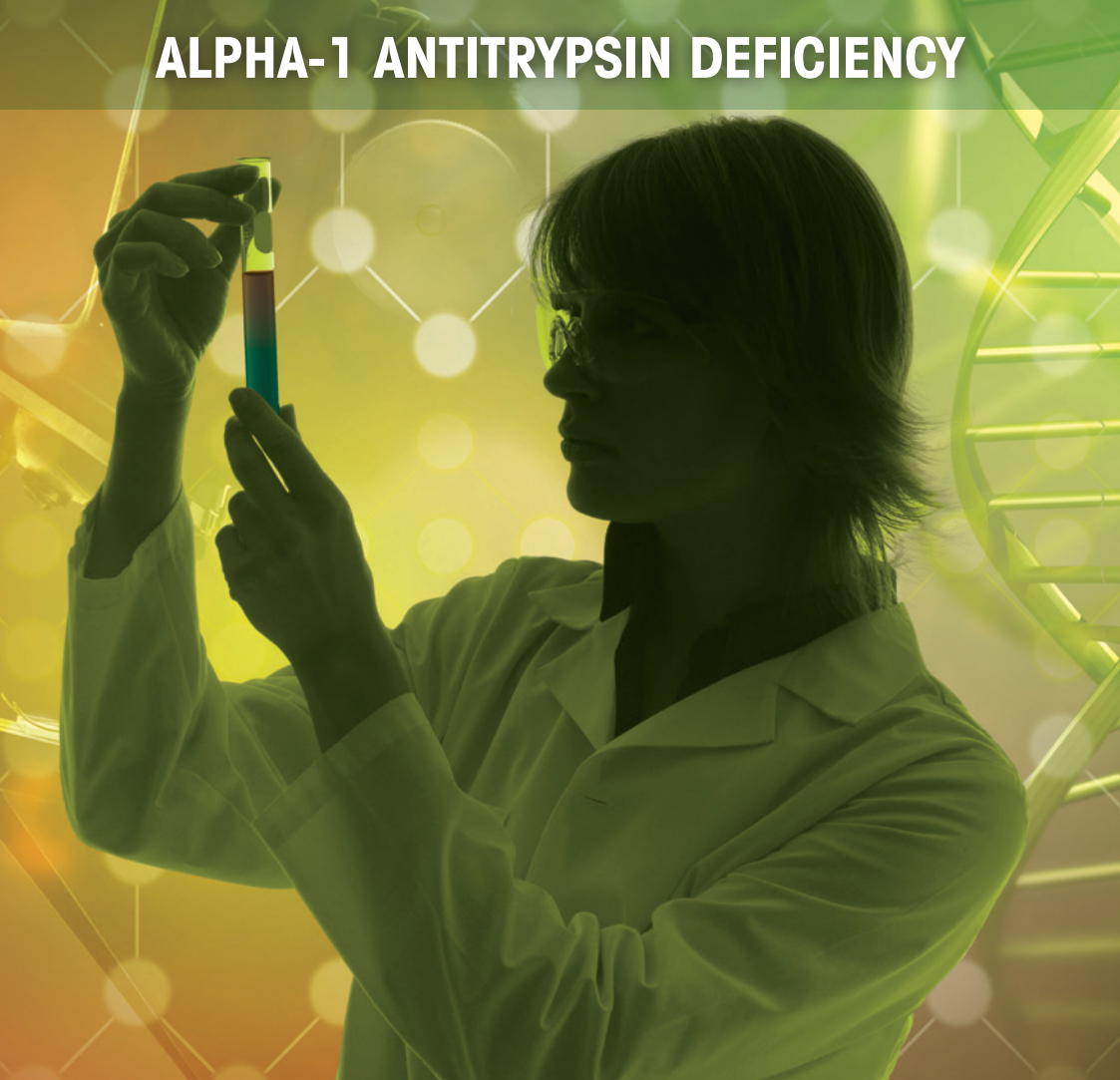




ALPHA-1 Association of Australia

**A GUIDE TO**

# **ALPHA-1 ANTITRYPSIN DEFICIENCY**





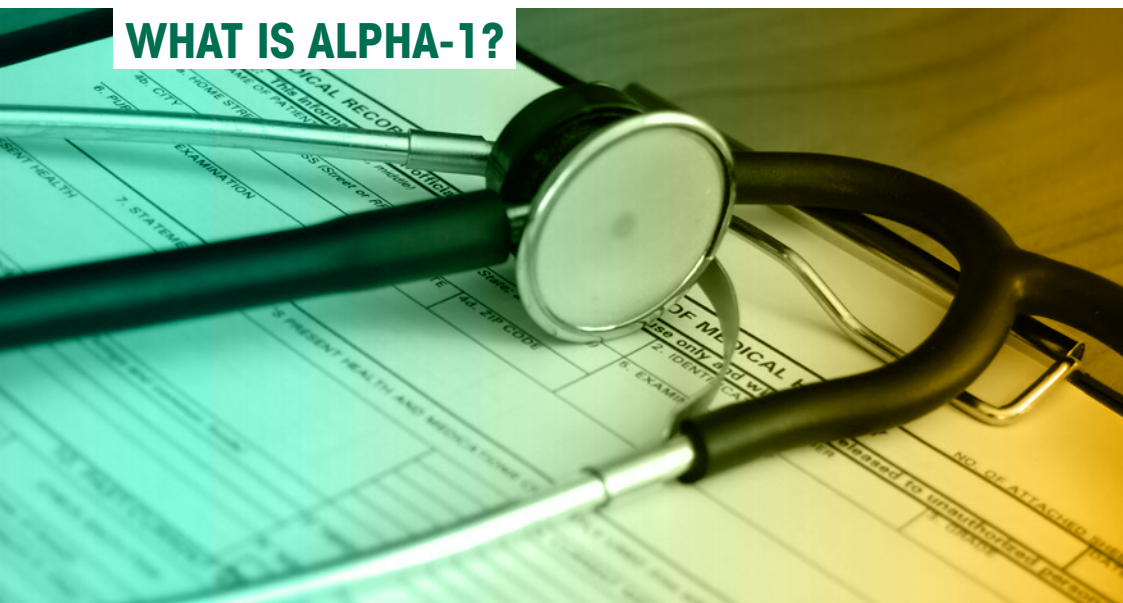
## DO YOU OR YOUR FAMILY MEMBERS SUFFER ANY OF THESE CONDITIONS?

- Asthma
- Jaundice
- Early onset emphysema
- Abnormal liver function
- Chronic bronchitis
- Recurrent chest colds
- Chronic Obstructive Pulmonary Disease (COPD)
- A family history of lung or liver issues

### **YOUR CONDITION COULD BE CAUSED BY AN HEREDITARY CONDITION KNOWN AS ALPHA-1 ANTITRYPSIN DEFICIENCY, OR ALPHA-1.**

The Alpha-1 Association of Australia (AAA) is an organisation dedicated to the help and support of Australians who are affected by and interested in Alpha-1.

# WHAT IS ALPHA-1?



Alpha-1 is a genetic disorder which can cause lung or liver disease in children and adults.

Alpha-1 is one of the most common hereditary disorders in the world, but it often remains undiagnosed.

Knowing that you have Alpha-1 gives you the opportunity to avoid risk factors and to make positive lifestyle changes which, together with the right medical care, can help to improve your quality of life.

Everyone's liver produces the protein alpha-1 antitrypsin, or AAT. An important function of AAT is to protect the lungs. In people with Alpha-1, the AAT is malformed and cannot be released by their liver. Alpha-1 occurs when there is a deficiency of AAT in the bloodstream.

AAT also protects delicate lung tissue from an enzyme known as neutrophil elastase. The normal function of neutrophil elastase is to remove inhaled material which has entered the respiratory system, such as smoke and other pollutants.

In the absence of AAT, neutrophil elastase will attack the harmful material as well as healthy lung tissue. Most commonly, this leads to lung disease, however, the build up of AAT in the liver can also lead to liver disease.

The deficiency can cause problems with the pancreas and is associated with the skin disease panniculitis. However, problems with the lungs, and to a lesser degree the liver, are far more common.

Not everyone who has Alpha-1 will develop symptoms. We still don't know why this is so, but we do know that early detection, treatment and lifestyle changes can make a dramatic difference in the progress of disease.

As a result, the sooner a diagnosis is made, the more effective lifestyle and treatment options will be.

## WHO IS AFFECTED BY ALPHA-1?



As a genetic condition, Alpha-1 is passed from parents to children through genes. Everyone has two AAT genes, both being normal in most people. Those who suffer symptoms of Alpha-1 may have either one normal and one defective gene (known as carriers), or two defective genes.

Approximately 1 in 37 worldwide have at least one defective gene\*. In Australia it is estimated to be about 1 in 9. The severity of the disease varies between individuals, as AAT levels in the bloodstream may range between zero-80% of normal levels, depending on the number and type of affected genes an individual has.

\* Estimate based on data from:  
de Serres (2002), Chest, v. 122, p 1818-1829

# WHAT ARE THE SIGNS AND SYMPTOMS OF ALPHA-1-RELATED DISEASE?

The World Health Organisation makes the following recommendation with regard to testing for Alpha-1:

**“It is therefore recommended that all patients with COPD and all adults and adolescents with asthma be screened once for AAT deficiency. Those with abnormal results on screening should have Pi (genetic) typing.”**

Alpha-1 affects people in different ways and symptoms can vary between individuals. Some common early symptoms associated with lung or liver dysfunction include:

- Shortness of breath
- Decreased exercise tolerance
- Recurrent chest colds or respiratory/lung infections
- Chronic coughing and/or wheezing
- Excess sputum (phlegm)
- Asthma which is not responsive to treatment
- History of suspected allergies
- Abdominal swelling
- Abnormal liver function tests or unexplained liver problems
- Enlarged liver or spleen
- Skin lesions or inflammation
- Jaundice



## HOW DO I GET TESTED FOR ALPHA-1?



If you suffer from any symptoms listed in this brochure, you can ask your doctor for a simple blood test which is covered by the Medicare Benefits Scheme. The Medicare schedule numbers for Alpha-1 testing are as follows:

### **66635 - Quantitation in serum, urine or other bodily fluids**

### **66638 - Isoelectric focussing or similar methods for determination of alpha-1 antitrypsin deficiency phenotype in serum**

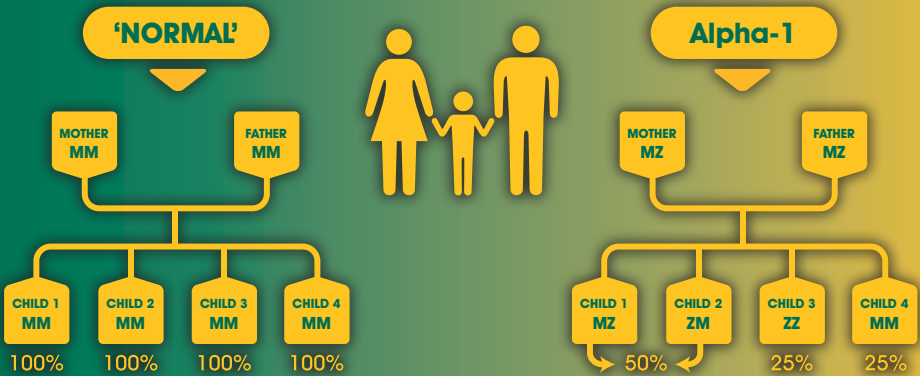
66635 will measure the level of AAT in your bloodstream. Low AAT levels may indicate a need for phenotyping (66638) or DNA testing to assess your individual AAT genetic makeup.

If you have a relative who has been diagnosed with Alpha-1, or a family history of early emphysema or liver disease, phenotyping or DNA testing is recommended regardless of your AAT levels. This is because AAT levels alone do not necessarily reveal your AAT genetic status. Knowledge of AAT levels in conjunction with knowledge of your AAT genetic status allows a more complete understanding of the implications for you and your children or future children.

Some with Alpha-1 may present without obvious symptoms and lead relatively healthy lives, especially if smoking is avoided. However, they still risk passing the defective gene on to their children.

Those seeking Alpha-1 diagnosis for themselves or family members should carefully consider health and life insurance complications, for example an insurer's requirement for disclosure of known pre-existing illnesses.

# HOW DID I INHERIT ALPHA-1?



Every human being is made up of many pairs of genes, and for each pair, we inherit one from each parent.

Pi stands for protease inhibitor, of which AAT is just one. A 'normal' person is referred to as PiMM, i.e. they have two normal genes, which means they produce a normal supply of AAT.

Sometimes a faulty gene is thrown into the mix. The most common abnormal gene is referred to as the Z gene (there are other variants, but since they all follow the same pattern, we'll stick with the Z). For someone to be PiZZ, a faulty gene must have been inherited from each parent, which means each parent must have had at least one Z gene (usually MZ).

See example in the diagram above.

Alpha-1 occurs when both parents pass on an abnormal gene to their child. A mother and father who are both carriers, (eg. MZ), have a 50% chance of their child being a carrier, 25% chance of having a child without Alpha-1, (MM), or 25% probability of having a child with Alpha-1, (eg. ZZ). The same percentages apply for each child. Therefore, it is possible for an MZ father and an MZ mother to have four children, all of who are ZZ or all of who are MM. On the other hand, it is possible for this same father and mother to have 1 ZZ child, 2 MZ children and 1 MM child, as the percentages imply. If one parent has the ZZ phenotype, and the other has two normal copies of the gene (MM), all of their children will have the MZ phenotype.

# CAN ALPHA-1 BE TREATED?



## TREATMENT OF ALPHA-1

Two manufacturers have registered their respective augmentation therapy products with Australia's Therapeutic Goods Administration. These therapies are Zemaira by CSL Behring, and ProLastin-C by Grifols. As at time of this booklet's production, the cost of augmentation therapy makes it out of reach for most Australian patients. The manufacturers have lodged applications for funding with the National Blood Authority, and the AAA eagerly awaits the outcome.

## TREATMENT OF SYMPTOMS

Treatment of the symptoms associated with Alpha-1 are many and varied, depending on the symptom. It is vital that the doctors involved with your care are aware of your Alpha-1 status, and the need to consider action at the first sign of related symptoms, for example bringing respiratory infections under control, in order to minimise long term damage.

## ORGAN TRANSPLANTATION AND OTHER SURGICAL PROCEDURES

Should your Alpha-1 lead to severe lung or liver damage, a lung or liver transplant, or lung reduction therapy, may be an option for you.



# WHAT IS MY FUTURE ONCE I AM DIAGNOSED WITH ALPHA-1?



Early diagnosis of Alpha-1 represents an opportunity to take steps to avoid risk factors, limit symptoms, or slow the progression of symptoms you may already be experiencing.

One of the things you should know is exactly which Alpha-1 genes you have. Without that knowledge, it is difficult to know how the diagnosis will affect either you or your family.

Not everyone with Alpha-1 will develop symptoms. Even if you do experience symptoms, proper treatment by your doctors and lifestyle changes may significantly increase the quality and quantity of your life.

# HOW CAN I IMPROVE MY CHANCES OF STAYING HEALTHY?



- Seek periodic liver and respiratory tests in order to help early identification of symptoms and to monitor progress
- Avoid cigarette smoking, fumes, dust, and other polluted environments, especially for extended periods, such as industrialised workplaces generating poor air quality
- Seek early treatment for lung infections and colds
- Seek your doctor's guidance in relation to vaccinating yourself against infectious diseases which may exacerbate the effects of Alpha-1
- Ensure you have regular immunisations against influenza and pneumonia
- Stay fit with regular exercise
- Maintain a balanced diet
- Moderate, if not eliminate, intake of liver toxins such as alcohol
- Avoid non-essential intake of drugs - some over-the-counter drugs and certain prescribed medications can harm the liver

# HOW CAN I HELP AAA?



To help the AAA meet its objectives, you can help in various ways:

## **Donations can be made via:**

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

## **Become a AAA volunteer:**

Contact us to find out about current projects, including fundraising.

## **Join the discussion:**

See the link to Discussions via

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

## **CONTACT DETAILS**

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