

For more information please visit:
www.alpha1.uk

or write to:
Alpha-1 Awareness
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Alpha-1 Awareness
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Alpha-1 Awareness

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Newly Diagnosed?

A guide for people diagnosed as having the
genetic condition Alpha-1 Antitrypsin Deficiency

www.alpha1.uk

Newly Diagnosed?

You are reading this as you have been told by your doctor that you have Alpha-1 Antitrypsin Deficiency or that the level of Alpha-1 Antitrypsin in your bloodstream is abnormally low. This leaflet attempts to explain, using the minimum of technical terms, what this means.

What is Alpha-1 Antitrypsin Deficiency?

Alpha-1 antitrypsin deficiency is a hereditary condition that occurs predominantly in people of European extraction. In the United Kingdom about one in 2,500 people have this condition. It is the most common genetic cause of pulmonary emphysema in adults and liver disease in children. In addition, there is evidence that infants, young children, and school-age children with the deficiency also have an increased likelihood of developing bronchial disease. According to current data, only around 2 to 5% of Alpha-1 patients are affected by liver disease in adult age.

History

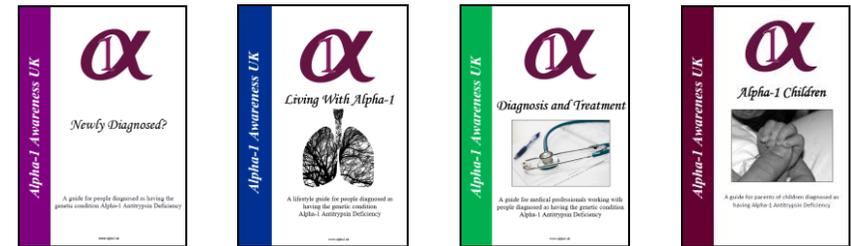
Alpha-1 antitrypsin deficiency (AATD) was first described as a hereditary disease by Laurell and Eriksson in 1963. Since its discovery our knowledge of the condition has expanded considerably, yet alpha-1 antitrypsin deficiency remains a largely unknown disease. People, like you, who have been diagnosed with the condition often have waited many years between the first appearance of symptoms and a correct diagnosis. In part this is because the symptoms often point to other, more common, complaints. The breathlessness that comes with chronic obstructive pulmonary disease (COPD) or signs such as jaundice, inflammatory liver disease and cirrhosis (scarring) of the liver are not always attributed to alpha-1 antitrypsin deficiency. The number of undetected Alphas is therefore agreed to be very high.

General therapeutic measures

Alpha-1 antitrypsin deficiency is not yet curable. At present only the symptoms can be treated by appropriate therapeutic measures. Besides specific treatments for the lungs and liver, all Alphas should take precautions to avoid infections. They should have a flu vaccination once a year and also receive immunisation against hepatitis A and B. **SMOKING** and alpha-1 antitrypsin deficiency are a deadly combination. For Alphas with liver disease the rule is **NO ALCOHOL**.

Our other publications

We created booklets for those who need more information about Alpha-1.



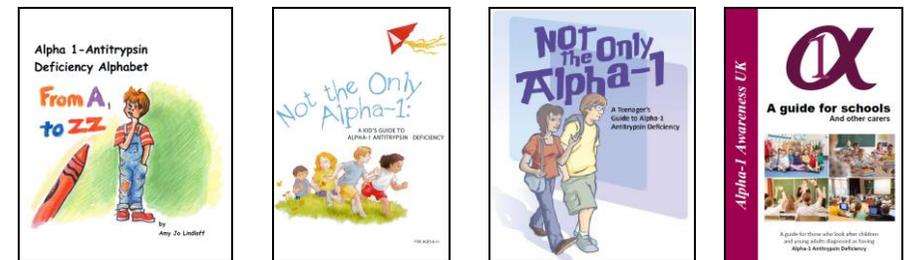
Newly Diagnosed – Information you need when you have just been diagnosed

Living with Alpha-1 – A lifestyle guide for Alphas

Your Child's Liver – For parents of an Alpha child

Diagnosis and Treatment – A guide for medical professionals working with Alphas

We also have booklets for the younger ones:



From A to ZZ – Designed for younger children 4 - 8

Not the Only Alpha-1 – For children aged between 8 – 11.

Not the Only Alpha-1 – For older children of 11 and up.

How to find us

Website

We have a web site full of news and information about Alpha-1. It is regularly updated with items that we feel will be of interest, from what medical trials are happening in the World, Study updates all the way to how to create your own cleaning products for your home (that have no harsh fumes or chemicals).

www.alpha1.uk

Facebook

We have a private Facebook Group for people connected to Alpha-1 to be able to talk to others about Alpha-1 and help support each other. You do not need to be a member of our charity to join the Facebook Group.

www.facebook.com/groups/alpha1awarenessuk

Post

If the internet is not your thing, you can send us a letter through the post.

Alpha-1 Awareness, PO Box 8294, Bakewell, DE45 9BF

Email

If you would like to email us, please use one of the below addresses. One of our Board members will then contact you as soon as they can.

General information - info@alpha1.uk

If you want to help fundraise for us - fundraising@alpha1.uk

For support - support@alpha1.uk

How does Alpha-1 Antitrypsin Deficiency develop?

Liver

Alpha-1 antitrypsin deficiency is a genetic defect that causes our liver either not to produce the protein called alpha-1 antitrypsin (AAT) or to produce it with errors in the structure of its molecules. These badly formed molecules fold incorrectly and become, by and large, unable to leave the liver. In contrast to the rest of the body, the problem in the liver is not a lack of but rather an accumulation of abnormally folded protein in the liver cells, and this damages them. In turn, the damaged cells can lead to prolonged jaundice in the newborn or bile stasis or chronic liver disease in children. Affected children can have poor appetite and fail to thrive. Although 10 to 20% of children with alpha-1 antitrypsin deficiency have liver involvement, that figure drops to 2 to 5% with increasing age. However, there are Alpha-1 patients who do not develop any symptoms until adult age. They are then more likely to develop cirrhosis (scarring) or cancer of the liver. As yet there is no specific drug treatment for liver Alphas.

Lungs

We inhale and exhale approximately 0.5 litres of air with every breath. This equates to around 10,000 litres a day. We also draw exhaust gases, dust, cigarette smoke (actively and passively), pollen, bacteria and much more into our lungs with every breath. The lungs defend themselves against these irritants with a substance (an enzyme) which is secreted by white blood cells and which keeps our lungs clean. Alpha-1 antitrypsin serves to stop this substance from attacking healthy lung tissue. If it is absent or in short supply, this protective function is insufficiently carried out, resulting in damage in the form of pulmonary emphysema, where the tiny air sacs are destroyed. The walls between the air sacs fuse, the lung tissue loses its elasticity and large cavities form and these fill with residual air. Shortness of breath develops – first during physical exertion, but in the advanced stage even at rest. Pulmonary emphysema is irreversible, i.e. the destruction of healthy lung tissue cannot be made good again. This process cannot as yet be halted, but it can be significantly slowed down by supplying alpha-1 antitrypsin in the form of weekly infusions (augmentation therapy).

In rare cases alpha-1 antitrypsin deficiency also damages other organs such as the skin (panniculitis or the inflammation of subcutaneous fatty tissue), the pancreas and blood vessels (vasculitis).

Who should be tested?

It is generally accepted that the following group of people should be test for AATD.

- All patients with frequently recurrent infections
- All patients with COPD (chronic obstructive pulmonary disease)
- All children, adolescents and adults with asthma
- Newborn, children and adults with liver disease of unknown origin
- All individuals in whose family alpha-1 antitrypsin deficiency has occurred

Since you have been diagnosed as having the genetic condition then it is very important that your close blood-relations are also tested for it. This way preventative measures against emphysema and liver scarring can start as soon as possible.

Testing for the deficiency can only be performed by medically qualified people and will probably be very similar to some of the tests performed on you (AAT level in a blood sample, phenotyping).

Signs of Alpha-1 Antitrypsin Deficiency

Provided that the condition is diagnosed early, the development of lung disease due to alpha-1 antitrypsin deficiency can be delayed for years or possibly prevented altogether. People should think of the possibility of alpha-1 antitrypsin deficiency if any of the following symptoms occur alone or together:

Lungs	Liver
Frequent infections	Jaundice
Frequent coughs, phlegm production	Dark urine
Shortness of breath	Distressing itching
Bronchitis	Failure to thrive
Asthma	Inflammation of the liver

Remember that these are just guidelines and only a medical doctor can give an authoritative diagnosis.

Liver & Lung Support Groups

We support everybody as much as we can. Our board members are not medical professionals, so are unable to offer medical advise. For medical questions, you always should speak to your GP or specialist.

Children

Liver



Children's Liver Disease Foundation

www.childliverdisease.org

Children's Liver Disease Foundation (CLDF) was formed in 1980 and is a unique national charity dedicated to taking action against the effects of all liver diseases of childhood. Based in Birmingham, CLDF is a comprehensive information hub for healthcare professionals and the general public, a tailored support service for children, young people and young adults with liver disease and their families.

Adults

Liver



British Liver Trust

www.britishlivertrust.org.uk

They support patients and families so you don't have to face liver disease alone. They campaign to improve awareness so more people are aware of the risks to the liver.

Lung



British Lung Foundation

www.blf.org.uk

They are the only UK charity looking after the nation's lungs. They have full support for lung affected people by phone, their online web community and also through their local support groups.

What can you do about it?

When first diagnosed most Alphas experience a turmoil of emotions and anger at the unfairness of life, guilt at passing on a defect to their children, fear of the unknown and many more. How they cope with this varies from person to person. One Alpha may use a particular stress management technique, another may find consolation in faith and others may simply accept it all.

The other thing that you can do is look at your lifestyle and see what changes may help you.

- Keep your body healthy with a balanced diet and regular exercise. Infections and other illnesses are more challenging to Alphas than to most people. Improve your defences.
- Smoking is especially harmful for people with AATD. Tobacco smoke releases increased amounts of the lung-damaging enzyme, thereby destroying more lung tissue. In addition, the smoke destroys what little AAT may be present in the lungs. If you smoke, you should quit as soon as possible. Certainly it is no easy matter to stop smoking, but you can do it. There are programmes to help you kick the habit and your GP will be able to advise you further. Even passive smoking is harmful. Ask everyone who smokes near you to refrain from doing so.
- Keep away from environmental pollutants such as open fires, petrol fumes, paints, solvents, even dust, etc. Some find room air fragrance sprays, deodorant sprays and hairsprays can also affect them.
- Heed any air quality warnings on the radio or television, especially in the hot summer months. If ozone levels are high, try not to venture outside and avoid excessive physical exertion.

Avoid contact with anyone who has a cold, the flu or chest infections as these can be very hard for an Alpha to get rid of.

Genotypes

Our ability to make AAT is inherited through genes passed on by both parents. We have about 25,000 pairs of genes in every cell of our body. Our genes determine how our bodies grow and develop. One of these genes is known as Protease Inhibitor (Pi). It is this gene that makes AAT.

We all have two copies of the Pi gene. We inherited one copy from our mother and the other from our father. When we have a child we pass one of our Pi genes, and our partner provides the other.

There are more than 75 different varieties of the Pi gene. Most of these varieties result in normal levels of AAT in the blood, but some result in low levels or no AAT. The most common varieties are labelled M, S and Z.

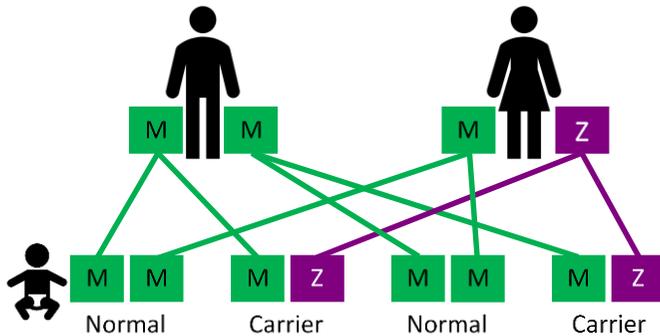
- Most people have two copies of type M (written PiMM) and have normal levels of AAT in their bloodstream.
- Type Z results in low levels of AAT in the blood stream and someone with two copies of the Z type (PiZZ) has AAT Deficiency.
- Someone with one copy of type Z and a copy of type M (PiMZ) is known as a carrier of AAT deficiency.
- Type S results in slightly reduced levels of AAT, so someone with PiMS will probably have less AAT than someone with PiMM but more than someone with PiMZ.

Your doctor may have arranged for various tests to be performed before you were informed of the diagnosis. The simplest of these is the level of AAT in the bloodstream. This can normally differentiate between Alphas and carriers of a faulty gene. More advanced laboratory tests look at the molecules of AAT and this can determine the types M, S, Z etc. This is called phenotyping. The most advanced tests analyse the Pi genes themselves. This complex and expensive process is called genotyping and it is used to find the extremely rare varieties of the Pi gene.

Heredity

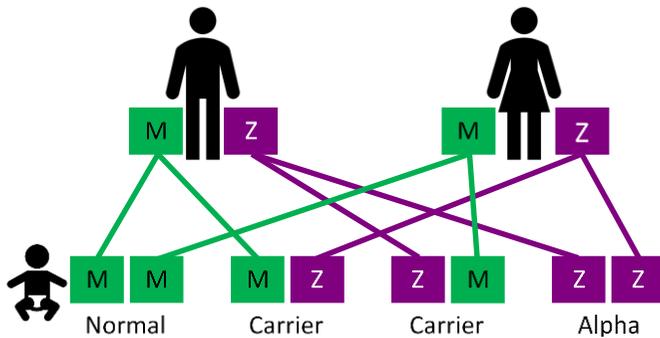
There are four ways of selecting two Pi genes, one from the mother's two Pi genes and one from the father's two Pi genes. Each child has just one of these four possibilities.

If you have been diagnosed as a carrier of AATD (PiMZ) and your partner is normal (that is to say PiMM), then the implications for your children are:-



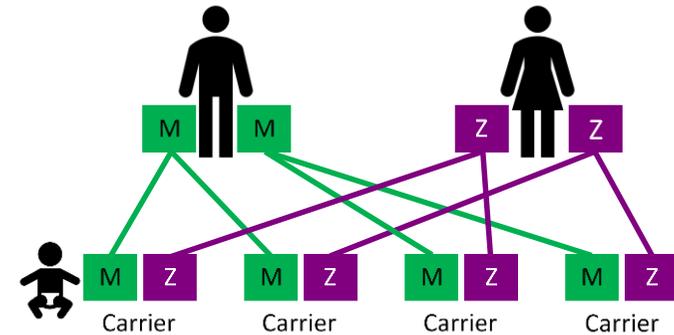
This shows an equal number of Carrier and Normal combinations and therefore each child will have a 50-50 chance of being a carrier.

If both parents are carriers (PiMZ), then there is a possibility of an Alpha child:



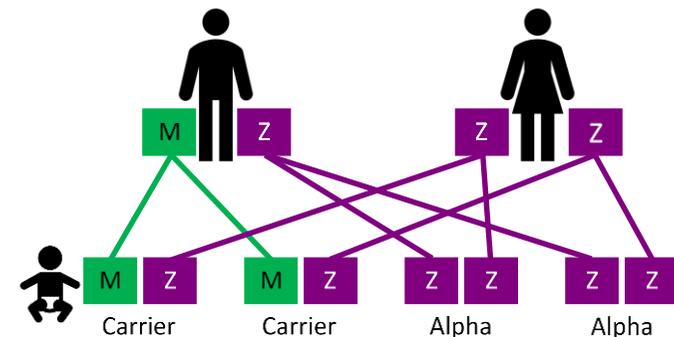
There is a 1 in 4 chance of being normal (PiMM), a 1 in 4 chance of being an Alpha (PiZZ) and there is a 2 in 4 chances of being a carrier (PiMZ).

If you have been diagnosed as being an Alpha (PiZZ) then your children will definitely be affected. If your partner is normal (PiMM) then the below are the potential outcomes:



Each possible outcome is that your child will be a carrier (PiMZ).

If your partner is a Carrier (PiMZ) there is more chances of an Alpha child:



There is a 1 in 2 chance the child will be a carrier (PiMZ) just as there is a 1 in 2 chance the child will be an Alpha (PiZZ).

If both parents are Alphas (PiZZ) then the children will also be Alphas.