

Could Your Asthma or COPD be Hereditary?



A Guide to Alpha 1 Antitrypsin Deficiency

Registered Charity England & Wales 1146330 : Scotland SC043177



Could your asthma or COPD be caused by an hereditary condition called alpha 1 antitrypsin deficiency

Alpha 1 antitrypsin deficiency, also called Alpha-1, A1AD or AATD, is a serious hereditary disorder which can result in lung or liver problems that can significantly impact on your life and even be life threatening.

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 (AAT). The job of AAT is to protect the body from inflammation, especially in the lungs. In people with Alpha-1 their AAT is malformed and cannot be released by their liver. Most commonly this leads to lung disease but the build up of AAT in the liver can also lead to liver disease.

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

While it is important to know that not everyone who has Alpha-1 will develop symptoms, even people who are "only carriers" sometimes do. 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.

There are many references to the Antitrypsin Deficiency Assessment & Programme for Treatment (ADAPT) throughout this booklet. This research programme is based in Birmingham and contact details can be found on the back cover.

How does lack of alpha 1 antitrypsin affect the body?

Lungs:

When someone gets an infection, the immune system kicks in and releases white blood cells into the bloodstream, these white cells attack and kill off the infection. As the white cells do their work, they release a potentially damaging enzyme called elastase. AAT prevents the elastase causing lung damage. To a lesser extent, the same thing happens throughout life, because white cells are constantly cleaning up inhaled germs and pollution.

Alpha-1 patients, whatever their gene combination, have less AAT in their bloodstream than normal. Those with the most serious gene combinations have hardly any. As a result, the elastase is able to cause damage, which it does by digesting healthy tissue. This damage occurs mainly in the lungs.

It is the loss of tissue over many years that results in serious lung disease. It can take 40 years or more for the damage to become noticeable.

Signs & symptoms of A1AD related lung disease

- Family history of lung disease.
- Rapid deterioration of lung function with or without a background of significant smoking, or occupational exposure to lung irritants.
- Asthma that is not fully responsive to treatment.
- Shortness of breath or awareness of ones breathing.
- Decreased exercise tolerance.
- Recurring respiratory infections.
- Chronic cough and sputum (phlegm) production (not always present).

Ask your doctor or contact ADAPT about being tested

Liver:

You now know that AAT is produced in the liver and that the AAT an Alpha-1 patient produces is abnormal.

Unfortunately as previously mentioned, this means that it gets stuck in the liver and cannot be released into the bloodstream. For most people this added complication doesn't cause many problems. However occasionally, the build up of the faulty AAT in the liver does cause damage, leading to liver disease.

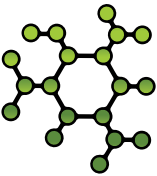
It isn't really understood why, but babies with Alpha-1 can occasionally be born with serious liver disease due to the build up of AAT during their development in the womb. So rarely, liver problems become apparent very early in life, however, most Alpha-1 patients will never have liver disease, with just a few experiencing symptoms later in life.

Signs & symptoms of A1AD related liver disease

- Family history of liver disease.
- Unexplained liver problems.
- Elevated liver enzymes.

Ask your doctor or contact ADAPT about being tested

How did I get alpha 1 antitrypsin deficiency?



As I'm sure you know, we are all made up of many pairs of genes and for each pair, we inherit one from each parent.

Pi stands for protease inhibitor of which alpha 1 antitrypsin is just one. A "normal" person is what is called PiMM i.e. they have two normal genes, this means that they produce a full supply of AAT. Sometimes a faulty gene is thrown into the mix; the most common abnormal gene is called 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 they must have inherited a faulty gene from each parent, which means in turn, that their parents must have had a least one Z gene (usually MZ).

How will this affect my own family?

If you have been diagnosed with one of the most severe genetic combinations, it does mean that your children will have inherited at least one faulty gene, because that is all you have to pass on. Unless your partner also has a faulty gene, your children will be carriers i.e. they will have one normal gene and one faulty gene. This is why many partners have blood tests themselves, to rule out a second faulty gene.



So, if you are ZZ or other serious combination, you will automatically pass on one faulty gene.

If your partner is also ZZ the children will be ZZ.

If your partner has just one faulty gene e.g. MZ, then the children could be either ZZ or MZ depending on which gene they have inherited from them.

If you are an MZ carrier and your partner is the same, then the children could be ZZ, if they are unfortunate enough to inherit both faulty genes. Alternatively, they could be MZ (or ZM, basically the same). On the other hand, they could be lucky and inherit both normal genes (MM).

It therefore follows that an MZ carrier partnered with someone who has normal MM genes, would have children who are either MZ carriers or completely normal, with MM genes.

Carriers of A1AD have less AAT than “normal” people but they do usually have enough in their bloodstream to prevent serious problems.

It isn't really understood why but all Alpha-1 patients, including some carriers, do seem to be more susceptible to picking up colds and 'flu, resulting in secondary chest infections. Therefore, it is just as important for carriers to look after themselves and it would be sensible to avoid smoking, and drinking to excess.

Testing for alpha 1 antitrypsin deficiency

Testing for AAT deficiency is simple and quick. It is usually done with a blood test. People at risk from alpha 1 antitrypsin deficiency should be tested. For more information about testing please visit our website: www.alpha1.org.uk or contact ADAPT who will be happy to help.



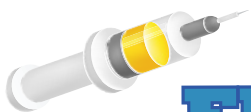
Who should be tested for AAT deficiency?

- The World Health Organisation (WHO), the American Thoracic Society, Alpha-1 Canada, the ADAPT Research Project and the Dept. of Health here in the UK, recommend that everyone with COPD be tested for alpha 1 antitrypsin deficiency.
- Everyone with emphysema, chronic obstructive pulmonary disease (COPD), chronic bronchitis or asthma that is not fully responsive to treatment.
- Individuals with bronchiectasis.
- Newborns, children and adults with unexplained liver disease.
- Individuals with a family history of liver or lung disease.
- Blood relatives of persons diagnosed with alpha 1 antitrypsin deficiency.
- Anyone with panniculitis, a skin disease.

Ask your doctor or contact ADAPT about being tested

How is alpha 1 antitrypsin deficiency treated?

Unfortunately, there is no specific treatment as yet for Alpha-1, so all that can be done is to treat the symptoms as they arise. It is vital that all the doctors involved in your care are aware of the deficiency, and the need to prescribe antibiotics and oral steroids at the first sign of a chest infection, to help prevent lung damage.



**Flu
Season**

In addition, it is very important to have the 'flu vaccination annually and also the pneumonia vaccination every five years.

The rest of your treatment will usually consist of various inhalers depending on which suit you the best.



Augmentation therapy, which replaces the missing protein through infusions of purified human AAT has been developed, but this is not yet available in the UK. However, there are ongoing trials of an inhaled form and it is hoped that this will eventually be approved and available, for some alphas at least, in the near future.

Your future with alpha 1 antitrypsin deficiency

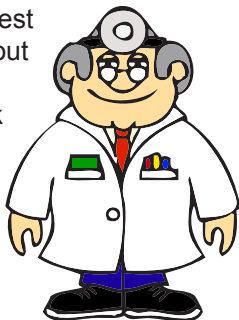
A positive diagnosis of alpha1 antitrypsin deficiency represents an opportunity to take concrete steps to avoid risk factors, limit symptoms, or slow the progression of symptoms you may already be experiencing.

Proper care by you and your doctors can have significant positive effects for the rest of your life.

Not everyone with Alpha-1 develops symptoms and even if you have, proper treatment by your doctors and lifestyle changes you can make, will significantly increase the quality and quantity of your life.

Therefore, even if you have developed symptoms, appropriate medication, treatments, exercise, and lifestyle choices, can go a long way toward ensuring you live a long and happy life.

Your doctor or consultant is the best person to provide information about the medications and treatment available for you now, and to look after you in medical terms, but there are important steps you can take to help take care of yourself.



It is important that you develop a good relationship with your doctors and consultants. Things are improving, but unfortunately, many are still not very knowledgeable about Alpha-1, so they may not always have the answers you are looking for. If that is the case, it would be advisable to ask for a referral to ADAPT.

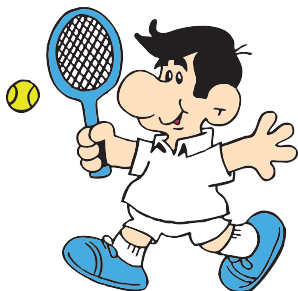
One of the first things you need to know is exactly which Alpha-1 genes you have, without that knowledge it is difficult to know how the diagnosis will affect either you or your family.

If you are symptom free, you have a good chance of remaining so by making small but important changes in the way you live.

Cigarette smoke is the greatest risk factor for developing life threatening symptoms. If you are a smoker, the single most important thing you can do to help yourself is to give up immediately. Passive smoking should also be avoided wherever possible.

Avoid other lung irritants, especially environmental pollutants used in agriculture, mineral dust, gas and fumes.

Regular exercise and good nutrition are beneficial in maintaining lung health, as is maintenance of the fat-soluble vitamins (A, D, E and K).



Eating a well balanced diet should provide good amounts of these fat soluble vitamins, although extra supplements may be advised by your doctor.

Remember, Alpha-1 is also a liver condition and fat soluble vitamins are stored in the liver and fatty tissues. Large doses of some vitamins can harm the liver.

Even if your symptoms are limited to respiratory problems, you also need to take care of your liver in order to minimize the possibility of liver disease.

Avoid things that may cause extra harm to the liver such as an excess of alcohol, street drugs, some over the counter drugs and certain prescribed medications. If in doubt contact your doctor.

There is no specific treatment for Alpha-1 associated liver disease. Eating a well balanced, healthy diet that contains plenty of fruits and vegetables, is essential to provide your liver with the antioxidants that it needs to protect against inflammation.



Researchers around the world are studying Alpha-1 and learning more all the time. Visit the Alpha-1 UK Support Group website regularly for the latest news and advice on managing your Alpha-1.

You can find us at: www.alpha1.org.uk



Join The Alpha-1 UK Support Group

Joining the support group is easy. Just visit our website: www.alpha1.org.uk and follow the “Mailing List” directions to the left of the screen. There is also a link to our Facebook group, if you would prefer to join us there.

- By joining us you will be in contact with a large community of Alpha-1 patients, their families and carers.
- We will keep you informed of all the latest news and information about Alpha-1, including new clinical trials.
- We will offer lifestyle tips to help you to keep as fit and active as possible.

If you don't have access to the Internet then you can contact us directly at the address on the back page. We can then arrange to send you an information pack and our six monthly newsletter.

Perhaps you could help raise funds to enable us to continue our work?



Whether £5 or £500, all donations will be put to good use, providing information, equipment and support for all Alpha-1 patients. In addition, we aim to promote better awareness and understanding of these A1AD related diseases throughout the medical profession.

For details of how to donate, or how to find us on JustGiving please visit our website.

It is also possible to raise funds without any cost to yourself, by using the [Easyfundraising](#) website to do your online shopping, or the [Easysearch](#) website for your internet searches. Again, please visit our website and follow the directions on the homepage to sign up to either of these free services.

The members of our support group face many issues that impact on the quality of their lives and the lives of other family members. Alpha-1 UK Support Group is committed to addressing these issues and to bringing about a well informed and responsive community. We have been supporting Alpha-1 patients for over fifteen years and we are backed by the UK's leading experts and researchers in the field of alpha 1 antitrypsin deficiency.

For more information about the Alpha-1 UK Support Group, ADAPT, or to help us with our work then please visit our website: www.alpha1.org.uk

Thank you.



Join our Alpha-1 community today at:

www.alpha1.org.uk

also find us on Facebook Groups:

alpha-1 uk Support Group - Alpha-1

Antitrypsin Deficiency (Est 1997)

With Special Thanks to :



www.alpha1canada.ca

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