NHS
Basildon and Thurrock University Hospitals
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Patient information

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This is a smokefree Trust. Smoking is not allowed in any of our hospital buildings or grounds.

The Trust will not tolerate aggression, intimidation or violence.

Alpha-1 antitrypsin deficiency

You may have been told that you or someone in your family has, or is a carrier of, a condition called alpha-1 antitrypsin deficiency.

What is alpha-1 antitrypsin and what does it do?

Alpha-1 antitrypsin (AAT) is a protein which circulates in the blood and helps protect the tissues of the body from being damaged by chemicals contained in white blood cells. These chemicals carry out important functions in the body, but can also have harmful effects. Alpha-1 antitrypsin helps provide protection against these harmful effects and if AAT levels are low, damage can occur to some tissues in the body. Cells in the lungs are the most likely to be affected, and there is a small chance of liver damage.

What is alpha-1 antitrypsin deficiency?

Alpha-1 antitrypsin deficiency is an inherited condition in which the body does not produce the correct amounts of AAT. This means that the AAT cannot work as efficiently as it should, leading to the possibility of lung or liver damage.

How does alpha-1 antitrypsin deficiency come about?

Alpha-1 antitrypsin deficiency occurs because of a fault in the alpha-1 antitrypsin gene. Genes are codes which tell our body how to make the different proteins required.

All our genes come in pairs, and we receive one copy of each gene from our mother and one from our father. Alpha-1 antitrypsin deficiency occurs only when a person has two faulty copies of the alpha-1 antitrypsin gene.

People who only have one faulty copy of the alpha-1 antitrypsin gene are called ‘carriers’ of the deficiency. Although their alpha-1 antitrypsin levels are lower than normal, they are not as low as in those who have the deficiency with two faulty copies of the gene. This does not usually cause any health problems.

How is the alpha-1 antitrypsin deficiency passed on?

Someone who has alpha-1 antitrypsin deficiency will have two faulty copies of the alpha-1 antitrypsin gene. Every time they have a child, they will pass on one of these faulty copies. Usually this will combine with a ‘normal’ working copy of the gene from the other parent. This means that all the children will be carriers, but this would not normally cause any ill effects.

For someone who is a carrier of alpha-1 antitrypsin deficiency, there is a 50/50 (heads or tails) chance of passing on the faulty gene or the working gene to each child.

If a child receives two faulty copies of the alpha-1 antitrypsin gene, it will have alpha-1 antitrypsin deficiency. This is important, because in a few cases, this alpha-1 antitrypsin deficiency can cause serious liver disease in early infancy. Anyone who has alpha-1 antitrypsin deficiency, or is a carrier of alpha-1 antitrypsin

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might wish to have their partner tested if they are planning to have children. If the partner is also a carrier, the couple can be given the opportunity to discuss the implications.

What does it mean for me if I have alpha-1 antitrypsin deficiency?

It is possible that you will have no ill-effects at all. Some people with this condition are completely unaware of it. It is possible that you may develop some problems with emphysema (lung disease). If you smoke you will certainly develop lung problems and these are likely to affect you by early middle age. If you do not smoke, problems are unlikely to arise until later life, if at all.

There is a small chance that you will develop liver problems. Your doctor might suggest some blood tests to check the function of your liver.

What does it mean for my children if I have alpha-1 antitrypsin deficiency?

If you have alpha-1 antitrypsin deficiency, all your children will be carriers. This will not affect their health unless they smoke (see below). If you plan to have more children, you might wish to arrange for your partner to be tested. This is because of the small chance of a child having a serious form of liver disease if both parents are carriers.

If your children are adults and planning to have their own children, they might wish to arrange to have their partners tested through their local GP.

What does it mean for me if I am a carrier of alpha-1 antitrypsin deficiency?

If you have been told you are a carrier of alpha-1 antitrypsin deficiency, this means that you have one faulty copy of the gene and one working copy. In this situation, there is very little chance of any serious health effects. The only exception to this is in smokers who are at greater risk of developing serious lung disease.

If you are planning to have children, you might wish to have your partner tested (see above).

What does it mean for my children if I am a carrier of alpha-1 antitrypsin deficiency?

If you are a carrier of alpha-1 antitrypsin deficiency, there is a 50/50 chance (heads or tails) chance for each child to be a carrier also. This will not affect their health. If your children are planning to have their own children, they might wish to arrange a test to see if they do carry alpha-1 antitrypsin deficiency. If that is the case, they can then consider arranging a test for their partner.

How can my relatives be tested for alpha-1 antitrypsin deficiency?

A simple blood test can be arranged through their family doctor.

Where can my relatives get advice?

The most important advice for any relative of someone who is a carrier, or affected with, alpha-1 antitrypsin deficiency is that they should not smoke. Non-smokers are most unlikely to develop serious problems associated with alpha-1 antitrypsin deficiency.

Anyone in the family who is already a smoker should consider giving it up. The family doctor should be able to give advice and support about this.

If a relative and his or her partner are both found to be carriers, and are planning children, they can ask for a referral to a genetic centre, where information can be given about the risks to any children, and options for management. Referral to a genetic centre can be made by the family doctor (GP), or by the hospital doctor who is already advising you.

If you need any more information, please the telephone the Consultant Biochemist on 01268 524 900 extension 3029 or 3025, or ask your GP.