Alpha 1 antitrypsin deficiency (AATD)

This fact sheet explains:

- What alpha 1 is
- Common symptoms
- Who is affected by alpha 1
- Diagnosis and treatment.

Alpha 1 (alpha 1 antitrypsin deficiency, or AATD) is a genetic condition (passed on to you from your parents' genes) that can result in serious lung disease in adults and lung or liver disease in babies, children and adults.

People with alpha 1 have no or very low levels of a protective enzyme inhibitor called alpha 1 antitrypsin (AAT) in their blood. An enzyme inhibitor is a chemical that controls enzymes, or proteins, in the body to make sure they're working as they should. AAT is made in the liver and released into the bloodstream and is particularly important in helping to protect the lungs from toxins that can cause lung damage.

Alpha 1 is damaging to the lungs and the liver, but in slightly different ways. In people whose lungs are affected, a lack of these protective enzymes means they are more at risk of a lung condition called chronic obstructive pulmonary disease (COPD), which is actually a group of conditions that include emphysema and bronchitis. This is a result of damage to the lungs from breathing in smoke and other toxins. In those whose liver is affected, it's thought that abnormal AAT proteins get 'stuck' and build up in the liver, causing damage over time. Children with alpha 1 may develop liver disease as they get older, and as adults, are at risk of developing cirrhosis (extensive scarring of the liver due to ongoing damage). This can change both the shape of the liver and its ability to function properly. It is more common in people over 50 and in people who have common risk factors for fatty liver, such as being overweight or obese, drinking harmful levels of alcohol (see guidelines below) or having diabetes. Ongoing damage can lead to liver disease and failure, meaning a liver transplant may be necessary.

What are the symptoms of alpha 1 (AATD)?

Common symptoms that there is a problem with the liver include:

- Jaundice (yellowing of the skin and eyes)
- Build-up of fluid in the abdomen (ascites) or legs (oedema)
- Fatigue
- Pain over the liver area (top right of the abdomen)
- Weight loss
- Pale or very dark stools (poo)
- Dark urine (wee)
- Itching.

Who gets alpha 1?

We all have two genes that 'instruct' the body to make alpha 1 antitrypsin (AAT). The gene which produces AAT in the normal way is called M. If you have two M genes, one from your mother and one from your father (MM), you'll have a normal level of ATT in your blood. These gene combinations are called genotypes.

If one of your parents has the gene that causes AAT deficiency, most commonly the Z gene (a rarer variation is the S gene), and this is passed on to you, you will have a combination of normal and abnormal genes (MZ, or MS, genotype). People with this combination have reduced levels of AAT, although this won't always cause health problems. People with the MZ, or MS, genotype are known as carriers – this means you can pass the Z gene onto your children. Carriers are also at greater risk of developing liver fibrosis if they have risk factors for fatty liver, such as being overweight or obese, or consume excess alcohol (current Government guidelines state that men and women should drink no more than 14 units of alcohol a week, with two or three consecutive alcohol-free days each week). Therefore, it is especially important for carriers to address these risk factors if they apply.

If two parents have the Z gene, and you inherit both (ZZ genotype), you'll have very low levels of AAT in your blood (10-20% of normal levels). In some people, this doesn't cause any problems. In others, it can cause the health conditions as described above. Children with alpha 1 may have problems with their liver in early childhood, although this is usually temporary and most have normal liver function by the time they are in their late teens. In newborn babies, alpha 1 can cause jaundice, characterised by yellowing of the skin and eyes, but this is generally managed safely and causes no lasting problems.

How is alpha 1 diagnosed?

Your doctor will organise blood tests to look for the genotypes that indicate you may have alpha 1, and the amount of AAT present in your blood. If alpha 1 is suspected, you may be sent for scans so that doctors can get a better look at the shape and condition of your liver. To see if any damage has occurred, you may be required to have a liver biopsy, whereby a very small sample of liver tissue is removed using a thin needle, to confirm the diagnosis and extent of liver damage.

How is alpha 1 treated?

There is no specific treatment for alpha 1 itself. The focus is on treating any liver damage present, unless liver disease is so advanced that transplantation is deemed necessary. Doctors will concentrate on monitoring the condition, managing symptoms as they arise and keeping you as healthy as possible by advising on how to reduce risk factors known to damage the liver, such as drinking too much alcohol or being overweight.

Useful contacts and support

alpha1.uk
A charity that supports people living with alpha 1, and their families and friends.

BRITISH LIVER TRUST

Contact details
Call us: 01425 481320
Helpline: 0800 652 7330
Email us: info@britishlivertrust.org.uk
Visit our website: britishlivertrust.org.uk
Write to us: British Liver Trust, 6 Dean Park Crescent, Bournemouth BH1 1HL

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