Pi SZ Alpha 1 Antitrypsin Deficiency and other rarer types

Introduction
This leaflet is written for people with alpha 1 antitrypsin deficiency (α1AD) of the Pi SZ type.

What is α1 antitrypsin (α1AT)?
α1AT is a chemical made in the liver that circulates in your bloodstream.

What is α1AD?
Individuals with α1AD have very low levels of α1AT in their bloodstream. Individuals with PiSZ α1AT also have low levels of α1AT in their blood, but not as low as people with two Z genes.

Why is α1AT important?
α1AT protects lung tissue from an enzyme (elastase) released by white blood cells. Elastase fights infection in the lungs. However if not tightly controlled by α1AT, elastase can attack healthy lung tissue.

What causes α1AD?
Our ability to make α1AT is inherited through genes passed on by both parents. One of these genes is known as Protease Inhibitor (Pi). It is this gene that makes the α1AT.

How is it inherited?
We all have two Pi genes. We inherited one gene from our mother and the other gene from our father. When a couple has a child they each pass on one of their Pi genes.

What are the different genetic types?
There are more than 100 different variants of the Pi gene. Most variants result in normal levels of α1AT in the blood, but some result in reduced levels or no α1AT. The most common variants are called M, S and Z.

Most people have two copies of type M variant (written as PiMM) and have normal levels of α1AT in the bloodstream.

Pi SZ
Type Z results in low levels of α1AT in the bloodstream. Type S results in slightly low levels. Someone with PiSZ has a low level.

How common is α1AD?
Around 1 in 2500 people in the UK have α1AD.
Most people with α1AD have two Z variants (PiZZ).
Around 1 in 10 people are carriers for the S or Z variant (4% (1 in 25) of Northern European population carry Z and 6% (1 in 17) carry S). Other types (I, F, Mmalton) are ever rarer.

Information for Patients

What are the effects of having α1AD?

The effects of the deficiency are varied: some people remain entirely healthy.
Lung disease: People who smoke and have PiSZ AAT deficiency may have an increased risk of developing lung problems such as emphysema (although this still remains uncertain). Non-smokers are not believed to be at a greater risk than normal of lung disease.

If lung disease is present symptoms include shortness of breath, wheezing and recurrent chest infections. It is recommended that everyone in the UK with Chronic Obstructive Pulmonary Disease (COPD) is tested for α1AD.

Liver problems: α1AT is made in the liver. In PiSZ patients, the α1AT accumulates in the liver and can cause problems, however most people do not develop serious liver problems.

Pi SZ AAT deficiency is not thought to be associated with liver health problems.

What can people with PiSZ α1AT do to look after their health?

- Avoid smoking and passive smoking. Smoking attracts white blood cells to the lungs, and speeds up the development of lung disease. Other lung irritants e.g. dust particles and air pollution
- Be referred to a specialist centre for α1AD for regular monitoring. See your GP for early treatment of lung infections or breathing problems or if you are concerned about any symptoms mentioned in this leaflet
- Drink alcohol only in moderation and exercise regularly

Other Pi variants

Some people inherit rarer AAT genes such as Mmalton, I or F. There is less information available about these genes.

Mmalton AAT has a tendency to accumulate in the liver (it forms “polymers”) and therefore can be associated with liver disease. If Mmalton is inherited with a Z gene (PiMmaltonZ) there is an increased risk of developing lung problems such as emphysema just like ZZ. For this reason it is very important to give up smoking if you have PiMmaltonZ AAT deficiency.

People who inherit both an I and Z gene (PiIZ) appear to be at increased risk of liver and lung disease compared to healthy people, there is not much information available at present about I AAT. However, it is still safer not to smoke if you have PiIZ AAT deficiency.

F AAT can be associated with normal levels of AAT in the blood, but this particular type of AAT does not work as well as normal AAT. People who inherit both F and Z genes (Pi FZ) also seem to be at an increased risk of getting lung problems such as emphysema. Again, it is important not to smoke if you have Pi FZ AAT deficiency.

Further information

Alpha-1 UK Support Group
Email: **info@alpha1.org.uk**
Web: **www.alpha1.org.uk**

Alpha-1 Awareness UK
www.facebook.com/groups/alpha1awareness.uk

Information for Patients

Please use the space below to write down any questions you may have and bring this with you to your next appointment.

*With thanks to the Clinical Genetics Unit,
Birmingham Women's NHS Foundation Trust

If you need more advice about alpha 1 antitrypsin deficiency please contact:

Alpha-1 Specialist NHS Service/ADAPT Project
Lung Function and Sleep Department
Level 0, Outpatients Area 3Queen Elizabeth Hospital Birmingham
Mindelsohn Way, Edgbaston
Birmingham B15 2GW

Tel: 0121 371 3885
Email: ADAPT@uhb.nhs.uk
Website: www.uhb.nhs.uk/services/respiratory-medicine/alpha-1-antitrypsin-deficiency-aatd.htm

If you require this information in another format, such as a different language, large print, braille or audio version please ask a member of staff or email patientexperience@uhb.nhs.uk.