



Pi ZZ Alpha 1 Antitrypsin Deficiency

An information leaflet for patients and families

Building healthier lives

UHB is a no smoking Trust

To see all of our current patient information leaflets please visit
www.uhb.nhs.uk/patient-information-leaflets.htm

Introduction

This leaflet is written for people with alpha 1 antitrypsin deficiency (α 1AD) of the Pi ZZ type.

What is α 1 antitrypsin (α 1AT)?

α 1AT is a chemical made in the liver that circulates in your bloodstream.

Why is α 1AT important?

α 1AT protects lung tissue from an enzyme (elastase) released by white blood cells. Elastase fights infection in the lungs. However if not tightly controlled by α 1AT, elastase can attack healthy lung tissue.

What is α 1AD?

Individuals with α 1AD have very low levels of α 1AT in their bloodstream.

What causes α 1AD?

Our ability to make α 1AT is inherited through genes passes on by both parents. One of these genes is known as Protease Inhibitor (Pi). It is this gene that makes the α 1AT.

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 α 1AT in the blood, but some result in reduced levels or no α 1AT. 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 α 1AT in the bloodstream.

Pi ZZ

Type Z results in low levels of α 1AT in the bloodstream. Someone with two Z variants (Pi ZZ) has α 1AD.

What are the effects of having α 1AD?

The effects of the deficiency are varied: some people remain entirely healthy.

Lung disease: Adults are at increased risk of developing breathing difficulties from disorders such as emphysema and chronic obstructive pulmonary disease (COPD), particularly if they smoke. Some symptoms include shortness of breath, wheezing and recurrent chest infections. It is recommended that everyone in the UK with COPD is tested for α 1AD.

Liver problems: α 1AT is made in the liver. In people with Pi ZZ the α 1AT accumulates in the liver and can cause problems, however most people do not develop serious liver problems. Some newborns with Pi ZZ develop jaundice or cholestasis and the vast majority recover. A smaller number of people develop liver problems after the age of 18.

Skin problems: α 1AT can also cause a rare skin problem, panniculitis, associated with painful red lumps and patches on the skin which can be treated with steroids.

What can people with α 1AT do to look after their health?

1. 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 certain chemicals should be avoided where possible.
2. Be referred to a specialist centre for α 1AD 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.
3. Drink alcohol only in moderation and exercise regularly.

How common is α 1AD?

Around 1 in 2500 people in the UK have α 1AD.

Most people with α 1AD have two Z variants (Pi ZZ). 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).

Is there a treatment for α 1AT deficiency?

General measures: If you have lung problems, your doctor may prescribe medicine to help your lungs function such as inhalers. If you have asthma, it is especially important to control it.

You should call your doctor at the first sign of a chest cold or other chest infection, since white blood cells come into the lungs to fight these infections. Your doctor may want to vaccinate you against influenza and pneumonia.

Your doctor may want to recommend an exercise plan, or pulmonary rehabilitation (support to help you breath). Many people are helped by involvement with support groups, such as the one at The Queen Elizabeth Hospital Birmingham.

Special treatment: Augmentation therapy is now possible, which boosts the level of α 1AT in your blood. While this treatment is potentially exciting, is not yet available on the NHS.

Augmentation must be given directly into a vein at regular intervals. Research continues to carry out careful clinical studies to determine whether or not augmentation is important in treatment.

What is augmentation?

Augmentation is a concentrated form of AAT purified from human blood. It is normally given once a week. It increases the AAT in your blood to levels that may help to protect your lungs. There is no form of the product that you can take by mouth.

Augmentation is not a cure, and it cannot reverse the lung damage that already exists. It does not treat or prevent liver problems.

The safety record with administration of augmentation has been excellent. In particular, there have been no reports of getting an infection from it. The part of the blood used to make augmentation has been rigorously tested to ensure that there is no risk from viruses such as hepatitis B.

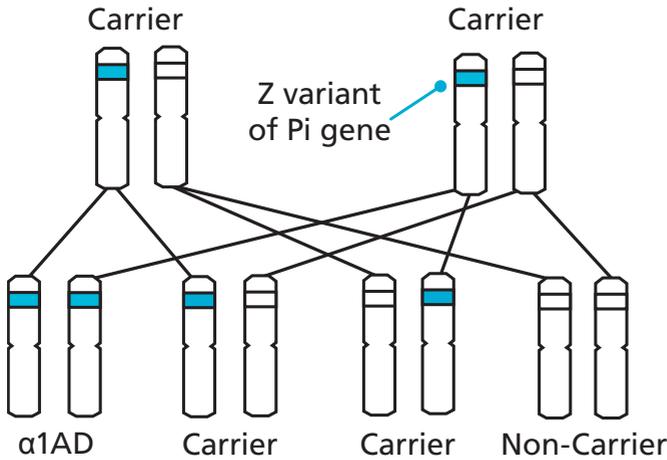
There are no exact alternatives to augmentation but non-specific treatments such as inhalers or (if necessary) lung transplant can be considered for any patient with emphysema (long term lung disease).

Will my children have α 1AD if I am a carrier?

If your partner is not a carrier you will not have a child with α 1AD. Your children will each have a 50% chance of being a carrier.

If your partner is also a carrier (Pi MZ) there will be three possible outcomes in each pregnancy.

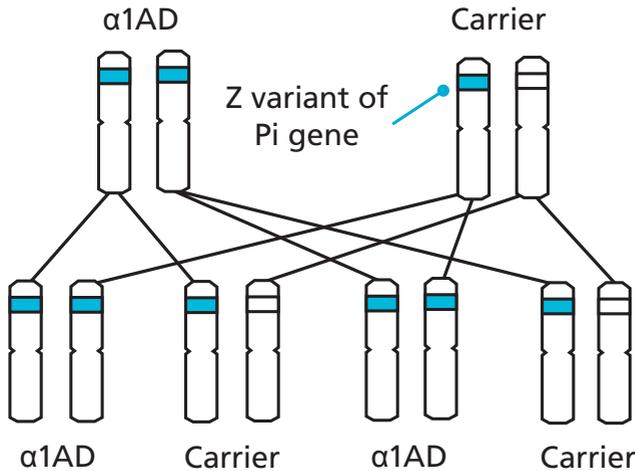
1. 1 in 4 (25%) chance you will both pass on the Z variant so your child will have α 1AD.
2. 1 in 2 (50%) chance one of you will pass the Z type and the other will pass the normal variant so the child will be a carrier of α 1AD.
3. 1 in 4 (25%) chance neither of you will pass on the Z variant so your child will not be a carrier of α 1AD.



Will my children have α1AD if I have it?

If your partner is not a carrier you will not have a child with α1AD. All your children will be carriers.

If your partner is a carrier your children will have a 50% chance of being a carrier and a 50% chance of having α1AD.



Not everyone with low levels of α1AD will develop significant health problems. Genetic testing will not indicate whether or not liver or lung problems will develop in the future.

Further information

Alpha-1 UK Support Group

36 Cecil Avenue, Lipson, Plymouth. PL4 8SG

Tel: 01752 225 573

Email: info@alpha1.org.uk

Web: www.alpha1.org.uk

Alpha-1 Awareness UK

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

Email: info@alpha1.uk

Web: www.alpha1.uk

*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 3
Queen Elizabeth Hospital Birmingham
Mindelsohn Way, Edgbaston
Birmingham B15 2GW

Tel: 0121 371 3885 or 3886 Fax: 0121 371 3887

Email: ADAPT@uhb.nhs.uk

Website: www.uhb.nhs.uk/alpha-1-antitrypsin-deficiency.htm



The Trust provides free monthly health talks on a variety of medical conditions and treatments. For more information visit www.uhb.nhs.uk/health-talks.htm or call 0121 371 4323.