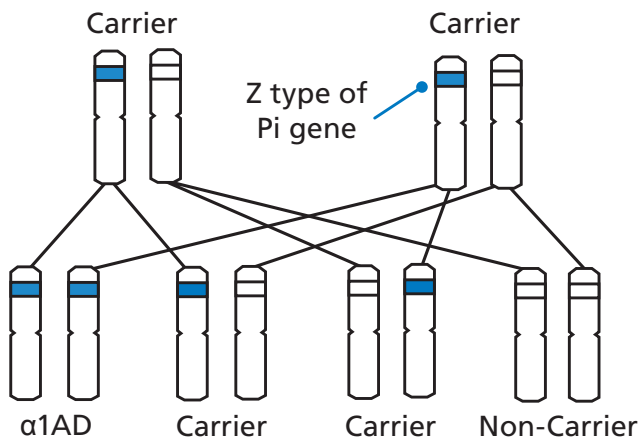


each have a 50% chance of being a carrier.

If your partner is also a carrier (PiMZ) there will be 3 possible outcomes.

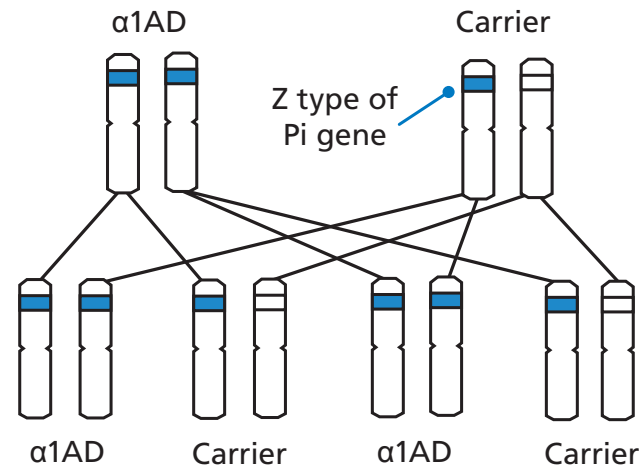
- 1) 1 in 4 (25%) chance you will both pass on the Z type so your child will have  $\alpha$ 1AD.
- 2) 1 in 2 (50%) chance one of you will pass the Z type and the other will pass the normal type so the child will be a carrier of  $\alpha$ 1AD.
- 3) 1 in 4 (25%) chance neither of you will pass on the Z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



### Will my children have $\alpha$ 1AD if I have it?

If your partner is not a carrier you will not have a child with  $\alpha$ 1AD. 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  $\alpha$ 1AD.



### Further information

Alpha-1 UK Support Group  
36 Cecil Avenue  
Lipson  
Plymouth, PL4 8SG  
Telephone: 0175 222 5573

*Birmingham Women's NHS Foundation Trust is not responsible for the third-party information and does not endorse any product, view or process or opinion from such sources.*

With thanks to the North East Thames Genetics Service, Great Ormond Street Hospital.

Birmingham Women's  
NHS Foundation Trust  
Edgbaston, Birmingham, B15 2TG  
Telephone: 0121 472 1377  
Fax: 0121 627 2602

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Birmingham Women's   
NHS Foundation Trust

# Alpha 1 Antitrypsin deficiency

An information leaflet for  
patients and families

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

**Clinical Genetics Unit**  
Birmingham Women's  
NHS Foundation Trust  
Mindelsohn Way  
Edgbaston  
Birmingham  
B15 2TG

Telephone: 0121 627 2630  
Fax: 0121 627 2618

Email: [Clinicalgenetics.info@bwhct.nhs.uk](mailto:Clinicalgenetics.info@bwhct.nhs.uk)

## Introduction

This leaflet is written for people with alpha 1 antitrypsin deficiency ( $\alpha$ 1AD) or people who are found to be carriers.

## What is $\alpha$ 1 antitrypsin ( $\alpha$ 1AT)?

$\alpha$ 1AT is a chemical made in the liver that circulates in your bloodstream.

## Why is $\alpha$ 1AT important?

$\alpha$ 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  $\alpha$ 1AT, elastase can attack healthy lung tissue.

## What is $\alpha$ 1AD?

Individuals with  $\alpha$ 1AD have very low levels of  $\alpha$ 1AT in their bloodstream.

## How common is $\alpha$ 1AD?

Around 1 in 2500 people in the UK have  $\alpha$ 1AD. 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).

## What are the effects of having $\alpha$ 1AD?

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

**Lung problems:** Adults are at increased risk of developing breathing difficulties from disorders such as emphysema and chronic obstructive pulmonary disease (COPD). It is recommended that everyone in the UK with COPD is tested for  $\alpha$ 1AD

**Liver Problems:**  $\alpha$ 1AT is made in the liver. A small proportion of individuals will develop liver problems in the neonatal period or later on in adulthood.

## What causes $\alpha$ 1AD?

Our ability to make  $\alpha$ 1AT 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  $\alpha$ 1AT.

## How is it inherited?

We all have two copies of the Pi gene. We inherited one copy from our mother and the other 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 70 different varieties of the Pi gene. Most varieties result in normal levels of  $\alpha$ 1AT in the blood, but some result in reduced levels or no  $\alpha$ 1AT. The most common varieties are called M, S and Z.

Most people have two copies of type M (written as **PiMM**) and have **normal** levels of  $\alpha$ 1AT in the bloodstream.

Type Z results in very low levels of  $\alpha$ 1AT in the blood stream. Someone with two copies of the Z type (**PiZZ**) has  $\alpha$ 1AD.

Someone with one copy of type Z and a copy of type M (**PiMZ**) is known as a **carrier** of  $\alpha$ 1AD.

Type S: Someone with PiMS will probably have less  $\alpha$ 1AT than someone with PiMM

but more than someone with PiMZ.

## What can people with $\alpha$ 1AD do to look after their health?

Anyone found to have  $\alpha$ 1AD should avoid smoking and passive smoking. Smoking attracts white blood cells to the lungs, and speeds up the development of lung disease. Similarly other lung irritants e.g. dust particles and certain chemicals should be avoided where possible.

A person with  $\alpha$ 1AD is more likely to develop liver problems. As with all individuals, they are advised not to exceed their weekly recommended intake of alcohol.

## What about people with PiSS

They will have slightly more  $\alpha$ 1AT in their bloodstream compared to people with PiZZ. They are therefore less likely to develop lung or liver problems. However they can follow advice given to people with PiZZ.

## Is there any special advice for carriers?

Being a carrier of  $\alpha$ 1AD does not normally have any effect on your health. However it is advisable to avoid cigarette smoke. It is thought that carriers who are smokers are slightly more likely to develop lung problems.

## Will my children have $\alpha$ 1AD if I am a carrier?

If your partner is not a carrier you will not have a child with  $\alpha$ 1AD. Your children will