



# Partial Alpha 1 Antitrypsin Deficiency (Pi MZ)

An information leaflet for patients and families

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## Introduction

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

## 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. Carriers have levels which are low but much closer to normal.

## How common is being a carrier of $\alpha$ 1AD?

Around 1 in 10 people are carriers for the S or Z variant. 4% (1 in 25) of the Northern European population carry Z and 6% (1 in 17) carry S.

## What are the effects of being a carrier of $\alpha$ 1AD?

All the information in studies carried out throughout the world indicates that having partial deficiency (Pi MZ) is not associated with any specific tendency to develop severe health problems. There has been some information to suggest there may be a very slight increased tendency to developing liver cirrhosis. There may be a possible slight relationship to the development of asthma, but these people are certainly no more likely to develop emphysema than any other healthy person who smokes.

## 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 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  $\alpha$ 1AT in the blood, but some result in reduced levels or no  $\alpha$ 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  $\alpha$ 1AT in the bloodstream.

### PiZZ

Type Z results in low levels of  $\alpha$ 1AT in the bloodstream. Someone with two Z variants (PiZZ) has  $\alpha$ 1AD.

## What do carriers of $\alpha$ 1AT need to 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. Ask family members or partners to get tested for  $\alpha$ 1AD.

The reason for doing this is chance. Everyone has a 3 to 6 chance in 100 of marrying or partnering another person with a similar partial deficiency, as it is quite common in the general population.

If we identify such a partner this has very important implications for their present or future children.

For instance, if a Pi Z deficient patient marries a partially deficient (Pi MZ) person, 1 in 2 of their children is likely to have severe deficiency (Pi Z) as well as their parent (the patient we originally found).

If on the other hand a person with partial alpha 1 antitrypsin deficiency (Pi MZ) marries another patient with partial deficiency, 1 in 4 of their children, again by chance, should develop severe deficiency of the Pi Z type.

It is the detection of these children that is the most important factor. By understanding that they have the deficiency, they can be advised with the help of their parents never to take up smoking and, if so, it is highly unlikely that they will develop severe emphysema. We know that prompt treatment of all chest problems is important in retaining the health of these children and also they require advice on which jobs would or would not be suitable for someone with their tendency to develop lung disease.

In other words, it is the aim of doing these tests to prevent ill health in children and grandchildren, nephews and nieces before it really starts.

## 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 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  $\alpha$ 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  $\alpha$ 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  $\alpha$ 1AD.







\*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:

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### **Alpha-1 Specialist NHS Service/ADAPT Project**

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