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.

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ADAPT

Antitrypsin Deficiency Assessment & Programme for Treatment

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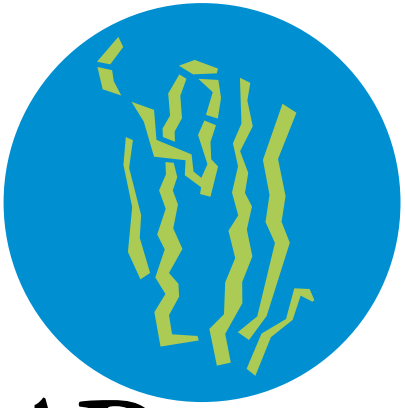
Partial Alpha1 Antitrypsin Deficiency (Pi MZ)



ADAPT

Severe alpha$_1$ antitrypsin deficiency is most commonly associated with two abnormal ZZ genes, each of which only makes one tenth of the alpha$_1$ antitrypsin you need. This is the type of deficiency that affects health, causing emphysema and cirrhosis of the liver.

In each family with a patient who has the Pi Z deficiency, we also find brothers and sisters with partial deficiency (Pi MZ), where one of the genes (M) is normal. These people make at least six-tenths of the alpha$_1$ antitrypsin they need. We are often asked what this means to the individual and what else needs to be done.

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 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 should we do?

For the person with partial deficiency we need to do very little unless incidental illness occurs, in which case it should be treated along the usual lines. However, we routinely screen the partners of our patients with severe Pi Z deficiency and those of their brothers and sisters who we identify as having partial (Pi MZ) deficiency.

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 are 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