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Partial Alpha-1-Antitrypsin Deficiency (PiSZ and other rarer types)
What is alpha 1 antitrypsin?

Alpha 1 antitrypsin (AAT) is a protein that is made in the liver and circulates in the blood. It is also called “alpha 1 proteinase inhibitor” by some scientists. AAT protects the tissues of your body (particularly the lungs) from being damaged by substances (enzymes) released from white blood cells. White blood cells release certain enzymes (called neutrophil elastase and proteinase 3) to help the cells move through tissues and to clear up infections. However, the activities of these enzymes must be strictly controlled, or they can attack normal tissues in your body. AAT normally provides one type of protection against these enzymes.

What is AAT deficiency?

Severe AAT deficiency is most commonly associated with inheriting two abnormal Z genes (PiZZ). Having two Z genes is associated with lung problems such as emphysema, and liver disease. Some individuals inherit one S gene from one parent and one Z gene from the other parent (PiSZ). Others may inherit rarer AAT genes such as Mmalton, I or F.

What do I need to know about PiSZ AAT deficiency?

Individuals with PiSZ AAT deficiency also have low levels of AAT in their blood, but not as low as people with two Z genes. 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). However, if you have any lung disease it is very important to give up smoking if you have Pi SZ AAT deficiency. Non-smokers are not believed to be at a greater risk than normal of lung disease. Pi SZ AAT deficiency is not thought to be associated with liver health problems.

If a person with PiSZ AAT deficiency has children with a partner who also carries an abnormal Z gene then there is a chance that their children could have severe AAT deficiency (Pi ZZ). Therefore people with PiSZ AAT deficiency who have or wish to have children should have their partner tested to find out whether they carry the Z gene. It is then important to identify children with PiZZ AAT deficiency early so that they can modify their lifestyle and be advised never to take up smoking. We know that prompt treatment of chest problems is important in retaining the health of these children, and they may require advice on which jobs would or would not be suitable for somebody with their tendency to develop lung disease.

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, but 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.

What information is available about other AAT genes?

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, but 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.