Melbourne Respiratory & Sleep Services

9459 0555 9455 0786
reception@mrss1.com.au
www.melbourne respiratoryandsleep.com.au
226 Burgundy Street Heidelberg VIC 3084

ALPHA-1 ANTITRYPSIN DEFICIENCY & LUNG DISEASE

Alpha-1-antitrypsin deficiency is an inherited disorder which usually affects the lungs. The information presented here is intended to answer your questions and provide you with a better understanding of this rare condition.

What is alpha-1-antitrypsin?

Alpha-1-antitrypsin is a protein which is produced by the liver and enters the blood stream. Its main role is to protect the lungs from destruction by other proteins called enzymes. Enzymes are found in all parts of the body and are needed for digestion to ensure that chemical reactions in the body proceed normally. Enzymes are also involved in areas of inflammation and tissue injury where they occur as a result of cell damage.

What is alpha-1-antitrypsin deficiency?

Alpha-1-antitrypsin deficiency is present when there is less than the normal amount of this protein in the blood. It becomes important only when the concentration in the blood is less than 20% to 30% of what we would normally expect. When this deficiency occurs, the lung is poorly protected from destructive enzymes and loss of lung tissue occurs, leading to a condition called emphysema.

One in every 2500 people in Australia has severe alpha-1-antitrypsin deficiency (levels below 20% of normal). This translates to nearly 7000 Australians with alpha-1-antitrypsin deficiency.

What is emphysema?

Emphysema is an irreversible condition which results from destruction of air sacs in the lungs. The air-sacs normally allow for absorption of oxygen into the blood and for the elimination of waste carbon dioxide. If they are destroyed, less oxygen enters the blood and breathlessness results. Cigarette smoking is the commonest cause of emphysema. Smoking makes the emphysema caused by alpha-1-antitrypsin deficiency much worse.

How will emphysema affect me?

The commonest symptom in emphysema is breathlessness which gets worse as the emphysema progresses.

Can I have an alpha-1-antitrypsin deficiency and not get emphysema?

Yes, different people have different amounts of the alpha-1-antitrypsin protein in their blood. Not all people will develop emphysema as the amount of alpha-1-antitrypsin present in most cases will be enough to protect the lung from destruction. Most, but not all people with levels less than 30% of normal will develop emphysema. Levels above 30% of normal seem to give adequate protection unless the person is a heavy smoker.

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Can I relieve the symptoms of my emphysema?

Some patients find that the use of inhaled bronchodilators (eg. Atrovent™, Bricanyl™, Respolin™, Ventolin™) help relieve their symptoms, especially the breathlessness. You should discuss this with your doctor. It is important to avoid prolonged exposure to dust, fumes and especially cigarette smoke. These pollutants can cause an acceleration in the rate at which lung tissue is destroyed.

Amounts and type of alpha-1-antitrypsin in your body

The amount of alpha-1-antitrypsin in your blood varies from individual to individual. The exact type of alpha-1-antitrypsin and its concentration in the blood is determined by genetic messages called phenotypes. Many different phenotypes have been described in humans, but only a very few actually cause significant alpha-1-antitrypsin deficiency.
Some examples of these are:

Phenotype:	alpha-1-antitrypsin concentration
MM	normal
MS	80% of normal
MZ	60% of normal
SZ	40% of normal
ZZ	10% of normal

The MM , MS and MZ phenotypes, which are quite common, are adequate to protect the lung against destruction. The ZZ phenotype, which is rare, is the phenotype usually associated with emphysema.

How did I inherit this deficiency?

You inherit one gene from each parent at birth from a choice of two. If one of your parents had normal levels of alpha-1-antitrypsin (ie. phenotype MM) and the other had very low levels (ie. phenotype ZZ) then your phenotype would be MZ (one gene from each parent) and your lungs would be protected. If both parents have the MM or ZZ phenotype then you would be either MM or ZZ.

Can I be treated for alpha-1-antitrypsin deficiency?

At the present time there is no specific treatment for alpha-1-antitrypsin deficiency although some drugs such as bronchodilators may be helpful. These should only be used on your doctor's advice. In the future, intravenous infusions of alpha-1-antitrypsin concentrate may be possible for some patients. Higher blood concentrations of alpha-1-antitrypsin would result and hopefully this would protect lung tissue from destruction. At present this treatment is not available in Australia and it is important to realise that this treatment has not yet been proven to be effective.

Please Note: This information is intended by The Australian Lung Foundation to be used as a guide only and is not an authoritative statement. Please consult your family doctor or specialist respiratory physician if you have further questions relating to the information provided here.