

Alpha-1 Antitrypsin Deficiency

Alpha-1 Antitrypsin Deficiency (Alpha-1) is a condition that is passed on from parents to their children through genes. This condition may result in serious lung and/or liver disease at various ages in life.

Alpha-1 antitrypsin is a protein that is produced mostly in the liver. Its primary function is to protect the lungs from neutrophil elastase. Neutrophil elastase is an enzyme that normally serves a useful purpose in lung tissue-it digests damaged or aging cells and bacteria to promote healing. However, if left unchecked, it will also attack healthy lung tissue. Alpha-1 antitrypsin, in sufficient amounts, will trap and destroy neutrophil elastase before it has a chance to begin damaging the delicate lung tissue. Consequently, if an individual doesn't have enough alpha-1 antitrypsin, the enzyme goes unchecked and attacks the lung.

Most people have two normal copies of the alpha-1 antitrypsin gene that make the protein. Some people may have one normal copy and one damaged copy of the gene; they are considered Alpha-1 Carriers. Individuals with two damaged copies of the gene have the severe deficiency of the alpha-1 antitrypsin protein and considered to have "Alpha-1" and are referred to as "Alphas".

Alpha-1 Carriers with only one abnormal gene can produce enough protein to stay healthy, especially if they do not smoke. However, people with two damaged copies of the gene can't produce enough alpha-1 antitrypsin, which can cause several conditions. They are often diagnosed with emphysema as their primary disease. Other common diagnoses include COPD (chronic obstructive pulmonary disease), asthma, chronic bronchitis, and bronchiectasis. Alphas are usually quite susceptible to lung infections. In the Alpha-1 patient, any of these conditions can cause further damage if they aren't treated right away.

Another disease that some Alpha patients develop is cirrhosis of the liver. This scarring of healthy liver tissue affects Alpha-1 infants, as well as 12% to 15% of adult Alphas. Unfortunately, there is no cure for cirrhosis of the liver, regardless of its cause. Cirrhosis can be managed as a chronic condition if caught early and protective steps are taken. Still, a liver transplant is currently the only option available for advanced disease.

More rarely, Alphas may also have a disease known as panniculitis. Panniculitis is an inflammation in the fatty tissue under the skin. It can occur in both children and adults.

Alphas and physicians regularly speak of a patient's "primary disease." This means the principal way the deficiency is manifested in a given patient, whether in the lungs, the liver, or the skin.

The only FDA-approved treatment for alpha-1 antitrypsin deficiency is augmentation therapy. Augmentation therapy consists of weekly IV infusions of alpha-1 antitrypsin derived from human plasma. It is used to increase the concentration of the protein in the blood and lungs. There are roughly 100 Minnesotans diagnosed with this rare disease.

SOURCE: Alpha-1 Association