

ALPHA-1 Awareness Month: November

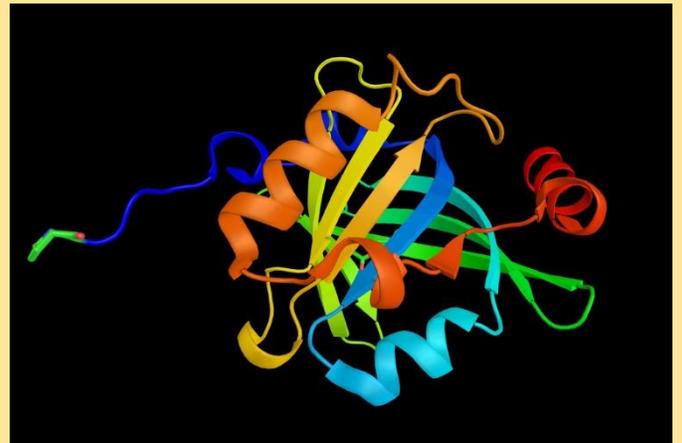
The Alpha-1 Association of Australia (AAA) was established for those with a genetically inherited medical condition which can affect the liver and lungs.

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.

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".



Since Alpha-1 Antitrypsin Deficiency is an inherited disorder, it occurs when both parents pass on a **abnormal gene** to their child. A father and mother who are both carriers (MZ) could expect to have a 50% chance of having a carrier, and a 25% chance of having either a healthy or a deficient child.

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.

Resource: www.alpha1.org.au

