**WHAT IS ALPHA-1?**

Alpha-1 is a **genetic inherited condition** which is passed from parents to children through genes.

Alpha-1 occurs when there is a **lack of a protein** in the blood called Alpha-1 antitrypsin, or AAT. AAT, the Alpha-1 protein, is **mainly produced by the liver**. The main function of AAT is to **protect the lungs from inflammation** caused by infection and inhaled irritants such as tobacco smoke.

The low level of AAT in the blood occurs because the AAT is abnormal and **cannot be released from the liver at the normal rate**. This leads to a build-up of abnormal AAT in the liver that can cause liver disease and a **decrease of AAT** in the blood that can lead to lung disease in adults.
**HOW IS ALPHA-1 INHERITED?**

As a **genetic disorder**, to inherit severe Alpha-1, a so-called **Z-AT gene** (or another severely impaired AT variant) must be passed on by both parents. If an individual carries two Z-AT, his/her level of AAT would be 10 to 20% of what it should be. The AAT protein affected by the Z-AT gene **builds up in the liver**, which means that the individual does not have enough AAT released to control the enzyme in his/her body. The defective production of AAT protein in the liver mainly results in **compromised pulmonary protection**.

**STANDARD OF CARE FOR ALPHA-1 PATIENTS**

Symptoms can be treated by **appropriate therapeutic measures**. Besides specific treatments for the lungs and liver, all Alpha-1 patients should take **precautions to avoid infections**, which includes **vaccinations**, **sensible consumption of alcohol** (unless liver disease is established in which case it should be avoided) and **no tobacco**.

Alpha-1 patients with **lung disease such as asthma, COPD or bronchiectasis** can be treated by the **same drugs that are used by non-Alpha-1 patients** for these conditions.

**Specific therapy** available to **Alpha-1 patients with lung disease** is called **augmentation therapy**. This therapy consists of intravenous infusions, usually weekly, of AAT protein in order to increase the level of the protein in the blood and lungs. The therapy has shown to reduce the rate of lung decline, and improve survival. The augmentation therapy cannot restore lost lung function, thus it is crucial for patients to receive it as early as possible if they have evidence of deteriorating lung function.

For **liver diseases**, **liver transplantation** is the **only viable therapy** today. Other treatment options may include adjustment of alcohol intake, hepatitis A&B vaccinations and nutrition adjustment.