ALPHA ONE ANTITRYPSIN CLINIC

The clinic will be providing blood test to screen for alpha one antitrypsin deficiency and assist patients who carry the disease to be placed on treatment.

What is alpha one antitrypsin A1AT?

A1AT is a protein protects the lung and the liver. Alpha-1 Antitrypsin Deficiency (Alpha-1) is a genetic (inherited) condition – it is passed from parents to their children through their genes. Alpha-1 may result in serious lung disease in adults and/or liver disease at any age.

For each trait a person inherits, there are usually two genes; one gene comes from each parent. People with Alpha-1 have received two abnormal alpha-1 antitrypsin genes. One of these abnormal genes came from their mother and one from their father.

The abnormal Alpha-1 genes.

There are many types of abnormal alpha-1 antitrypsin genes. The most common abnormal genes are called $S$ and $Z$. Normal genes are called $M$. A person who does not have Alpha-1 will have two M genes (MM).

The main function of AAT is to protect the lungs from inflammation caused by infection and inhaled irritants such as tobacco smoke.

The patients with alpha one deficiency are at risk to develop lung disease called emphysema and COPD.

The most common signs and symptoms of lung disease caused by Alpha-1

1. Shortness of breath
2. Wheezing
3. Chronic bronchitis, which is cough and sputum (phlegm) production that lasts for a long time
4. Recurring chest colds
5. Less exercise tolerance
6. Asthma that can’t be completely reversed with aggressive medical treatment
7. Year-round allergies
8. Bronchiectasis

**Alpha-1 cannot be diagnosed by symptoms or by a medical examination alone; you need to get a simple blood test to know for sure.**

- Alpha-1 is the most common known genetic risk factor for emphysema.
- Up to 3% of all people diagnosed with COPD may have undetected Alpha-1.
- Alpha-1 can also lead to liver disease. The most serious liver diseases are cirrhosis and liver cancer.
- The World Health Organization (WHO), American Thoracic Society (ATS), and the European Respiratory Society (ERS) recommend that **everyone with COPD be tested for Alpha-1.**

**Testing for alpha one:**

It is simple blood test. The test is finger puncture; we will place the drops of blood on special paper. The test will be provided free in the clinic. The test is done to evaluate the serum level of A1AT. Depending on the serum level further test can be done. If serum level is normal usually no further test is needed unless there is indication. If the serum level is abnormal then further tests need to be done. Usually we will do phenotype to look for abnormal gene.

Some patient with normal A1AT may still need to have phenotype evolution depending on clinical assessment.

**What is the treatment for alpha one deficiency?**

The specific therapy for the treatment of Alpha-1-related lung disease is augmentation therapy – also called replacement therapy. Augmentation therapy is the use of alpha-1 antitrypsin protein (AAT) from the blood plasma of healthy human donors to augment (increase) the alpha-1 levels circulating in the blood and lungs of Alphas diagnosed with emphysema. The therapy is administered by a weekly intravenous infusion and, until other therapies become available, it is considered ongoing and lifelong.

**What are the goals of therapy?**
The basic goal of augmentation therapy is to increase the level of alpha-1 protein in the lungs. Alpha-1 antitrypsin protects the lungs from the destructive effects of neutrophil elastase, an enzyme released by our body’s white blood cells as they respond to inflammation or infection.

The ultimate goal is to slow or stop the progression of lung destruction by replacing the deficient protein. The therapy cannot restore lost lung function — nor is it considered a cure. There is also some evidence that augmentation therapy can reduce the frequency and severity of pulmonary exacerbations (flare-ups of lung disease) and it appears to be an effective treatment for the Alpha-1 related skin disease, Necrotizing Panniculitis.

Who should receive augmentation therapy and when should they start?

Augmentation therapy should be given to individuals with documented emphysema and severe Alpha-1 (defined as individuals with two abnormal alpha-1 genes). There has been some controversy, however, about giving augmentation therapy to anyone whose lung disease is very mild or very severe.

In some research studies conducted since the introduction of augmentation therapy, the benefits identified were primarily seen in certain groups of people. In these studies, the most dramatic effects were seen in those people whose lung function fell in the mid-range of severity.

Things to consider before beginning augmentation therapy

Before beginning augmentation therapy, a patient should be tested for IgA deficiency, a hereditary condition that makes potentially severe allergic reactions to plasma products more likely.

AlphaNet recommends that immunization against both Hepatitis A and B be considered for all Alphas to reduce the risk of liver injury. Vaccination entails a series of three injections, generally administered over six months. Augmentation therapy can be started independent of whether or when hepatitis vaccine will be given.

The purpose of augmentation therapy

Augmentation therapy is intended to augment (add to) the amount of alpha-1 antitrypsin protein (AAT) floating in the blood and bathing the tissues of the body in people with lung disease related to Alpha-1. Although some people report that they notice improvements in their health when on augmentation therapy, and there is some evidence for a decrease in the number of lung infections in individuals.
receiving augmentation therapy, the primary aim of this therapy is to reduce the rate of decline of lung function towards normal and, therefore, improve the long-term quality of life and even the lifespan of those with lung disease due to Alpha-1.

Everyone loses lung function during their adult life, whether they have Alpha-1 or not. Alphas with lung disease lose their lung function at a more rapid rate than normal. If augmentation therapy is effective, it will be expected to slow this increased rate of decline, regardless of the severity of the underlying lung disease.

**Alpha one clinic will provide:**

1. Provide the test to evaluate for alpha one deficiency. The test is provided, no charge.
2. The clinic will help educate the patient about alpha one.
3. The clinic will communicate with the primary care physician about the results and if there is need to start treatment.
4. The clinic will help to start treatment for patient with alpha one.
5. The clinic will ensure communication with primary care provider on regular base to keep the primary updated on the treatment.

Resource, alpha one foundation.