ALPHA-1 FOUNDATION

Frequently Asked Questions about Alpha-1 Antitrypsin Deficiency

What is Alpha-1?

Alpha-1 Antitrypsin Deficiency (Alpha-1) is a genetic (inherited) condition that may result in serious lung disease in adults and/or liver disease at any age. Alpha-1 results in abnormal alpha-1 proteins that are mainly produced by the liver.

How does a person develop Alpha-1?

Alpha-1 is inherited – passed on from parents to their children through their genes.

What are the health consequences of Alpha-1?

People with Alpha-1 have an increased risk of developing emphysema (a form of COPD) and liver disease. Alpha-1 is the most common known genetic risk factor for emphysema. Of the more than 12 million people diagnosed with COPD in the U.S., up to 3% of them may have Alpha-1. Alpha-1 is commonly referred to as “genetic COPD.” The most serious liver diseases related to Alpha-1 are cirrhosis and liver cancer.

Who are affected and how many people have Alpha-1?

Alpha-1 affects men and women and has been identified in all races and ethnicities. It affects about 1 out every 2,500 people in the U.S., at least 100,000 people. About 10-15% of all liver transplant candidates have Alpha-1-related genetic abnormalities. Alpha-1 is one of the leading reasons for liver transplantation in children. An estimated 19 million people in the U.S. have one normal and one defective gene and are called carriers. Carriers may pass the defective gene to their children.

How is Alpha-1 diagnosed?

Alpha-1 can be diagnosed by a simple blood test. However, current data suggest that less than 10% of people living with Alpha-1 in the U.S. have been properly diagnosed. It takes an average of three doctors and seven years from the time lung symptoms first appear before proper diagnosis is made. Alpha-1 is often misdiagnosed as asthma or smoking-related COPD. Common symptoms include: shortness of breath, wheezing, coughing with or without sputum (phlegm) production, recurring respiratory infections, rapid deterioration of lung function, or unexplained liver disease and /or elevated liver enzymes. Early diagnosis can prevent life-threatening complications of Alpha-1.

ALPHA-1 FOUNDATION

Frequently Asked Questions about Alpha-1 Antitrypsin Deficiency

How are the lungs affected?

The abnormal alpha-1 protein is trapped in the liver, causing a deficiency of the protein that would normally circulate in the blood and protect the lungs from many types of damage. This can result in emphysema (holes in the lungs) even in people who have never smoked. Smoking can accelerate this type of damage. Despite treatments, including augmentation therapy, adults may require a lung transplant due to severe emphysema.

How is the liver affected?

Liver disorders are caused by the accumulation of the abnormal alpha-1 protein within the liver cells. The effects of such accumulation can range from abnormal liver function tests without symptoms to severe scarring (cirrhosis of the liver) and, rarely, liver cancer. Patients may require a liver transplant if the liver is severely affected.

Are there treatments specifically for people with Alpha-1?

The FDA has approved four plasma-derived augmentation therapy drugs in the U.S. specifically for people with lung disease due to Alpha-1. This may slow the progression of Alpha-1 lung disease; it cannot reverse emphysema. The only treatments for Alpha-1 liver disease are the usual therapies provided to everyone with significant liver disease, including liver transplantation.

Are there programs to provide support for patients and families?

The Alpha-1 Foundation has a Patient Information Line: 800-245-6809 and a network of local support groups throughout the U.S., as well as virtual support groups to help people and their families affected by Alpha-1. Other support programs include Alpha-1 Kids, Genetic Counseling services, A1F Education Days throughout the U.S., and an annual A1F National Conference.

Is there research under way on Alpha-1?

Yes. The Alpha-1 Foundation is committed to finding a cure for Alpha-1 and to improving the lives of people affected by Alpha-1 worldwide. It has invested over $100 million to support Alpha-1 research and programs at 130 institutions in North America, Europe, the Middle East, and Australia.

Why is there a need for public awareness of Alpha-1?

Alpha-1 is a common yet under-recognized condition. The public may not understand the symptoms, the need for testing and the potentially life-threatening consequences of Alpha-1, demonstrating the need for increased awareness.