Common Questions and Answers about Alpha-1 Antitrypsin Deficiency (Alpha-1)

Q: What is Alpha-1?
A: Alpha-1 Antitrypsin Deficiency or Alpha-1 is a genetic (inherited) condition that may result in serious, *chronic* lung and/or liver disease at various ages in life (children and adults). This genetic condition results in abnormal alpha-1 proteins that are mainly produced by the liver and secreted into the blood stream. It is the most common known genetic risk factor for Chronic Obstructive Pulmonary Disease (COPD). Of the more than 12 million people diagnosed with COPD in the US, and about 3 percent of them are predicted to have Alpha-1

Q: How does a person develop Alpha-1?
A: Alpha-1 is passed on from parents to their children through their genes.

Q: What are the health consequences of Alpha-1?
A: People with Alpha-1 have an increased risk for developing emphysema (a form of COPD).
The most serious liver diseases related to Alpha-1 are cirrhosis and liver cancer. About 10-15% of individuals with Alpha-1 are children affected by liver disease at birth.

Q: Who are affected and how many people have Alpha-1?
A: Alpha-1 equally affects men and women and has been identified in all races and ethnicities. that Alpha-1 affects 1 out every 3,000 people in the US, or 100,000 people.
About 10 to 15% of all liver transplant candidates have the Alpha-1 related genetic abnormalities. Alpha-1 is one of the leading reasons for liver transplantation in children.
An estimated 20 million people are considered Alpha-1 carriers, having one normal and one defection gene. Usually, in such individuals the protein blood levels may be normal or slightly reduced. Carriers may pass the defective gene along to their children.

Q: What can you tell me about the diagnosis of Alpha-1?
A: Alpha-1 can be diagnosed by a simple blood test. However, current data suggest that less than 10%, of those people living with Alpha-1 in the US 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 difficult to diagnose because of common respiratory symptoms and/or unexplained cirrhosis of the liver. Common symptoms of Alpha-1 include:

- A family history of Alpha-1, lung or liver disease.
- Shortness of breath
- Wheezing or non-responsive asthma
- Coughing with or without sputum (phlegm) production
- Recurring respiratory infections
- Rapid deterioration of lung function
- Unexplained liver problems and /or elevated liver enzymes

Early diagnosis saves lives and can prevent life threatening complications of Alpha-1.

Q: How are the lungs affected?
A: 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. The main result may be emphysema (destruction of supporting structures/air sacs within the lung), in younger adults (in their 30s and 40s), even in those who have never smoked. Smoking can accelerate this type of damage. Despite treatments, including protein replacement, adults may require a lung transplant due to severe emphysema.

Q: How is the liver affected?
A: 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, symptomatic scarring (cirrhosis of the liver) and, rarely, liver cancer. Children and adults may require a liver transplant if the liver is severely affected.

Q: Are there treatments specifically for people with Alpha-1?
A: Lung: The FDA has approved three plasma-derived augmentation therapy drugs in the US specifically for people with lung disease due to Alpha-1. Evidence suggests this may slow the progression of Alpha-1 lung disease; it does not cure the emphysema..

Liver: The only treatments for Alpha-1 liver disease are the usual therapies provided to all individuals with significant liver disease, including liver transplantation.

Q: Is there research underway in the field of Alpha-1?
A: Yes. The Alpha-1 Foundation, organized in 1995, is dedicated to providing the leadership and resources that will result in increased research, improved health, worldwide detection, and a cure for Alpha-1 Antitrypsin Deficiency. The Foundation has invested $35 million to support Alpha-1 Antitrypsin (AAT) research and programs in nearly 70 institutions in North America and Europe For more information, visit: www.alphaone.org

Q: Why is there a need for public awareness of Alpha-1?
A: Alpha-1 is a common yet under-recognized condition. The public may not understand the importance of selected symptoms and potentially life-threatening consequences of Alpha-1, demonstrating the need for increased awareness.
Source: Alpha-1 Association www.alpha1.org
Alpha-1 Foundation www.alphaone.org
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