

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

In the United States there are at least 100,000 people with Alpha-1 (ZZ).

The most common signs and symptoms of Alpha-1 are:

- Shortness of breath
- Wheezing
- Chronic cough and sputum (phlegm) production (chronic bronchitis)
- Recurring chest colds
- Decreased exercise tolerance
- Bronchiectasis
- Unexplained liver disease or elevated liver enzymes
- Eyes and skin turning yellow (jaundice)
- Swelling of the abdomen (ascites)

Alpha-1 has been identified in virtually all populations. About 1 in every 2,500 Americans has Alpha-1.

An estimated 19 million people in the U.S. have one normal and one defective alpha-1 gene. People with one normal gene and one defective gene (for example MZ) are called “carriers”. Carriers may pass the defective gene on to their children.

Alpha-1 can lead to lung destruction and is often misdiagnosed as asthma or smoking-related Chronic Obstructive Pulmonary Disease (COPD).

Alpha-1 is the most common known genetic risk factor for emphysema.

The American Thoracic Society (ATS) and the European Respiratory Society (ERS) recommend that everyone diagnosed with COPD, emphysema, or asthma that is not completely reversible with aggressive treatment, be tested for Alpha-1.

Alpha-1 can lead to liver disease. The most serious liver diseases are cirrhosis and liver cancer.

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

For more information, call toll-free: 1-877-228-7321 or visit www.alpha1.org.