



What you need to know about...

Alpha-1 Antitrypsin (AAT) Deficiency

What is Alpha-1 Antitrypsin Deficiency?

Alpha-1 Antitrypsin (AAT) Deficiency is a hereditary condition which causes your body to lack or produce low levels of this protective protein. AAT helps protect the lungs from the damaging effects of enzymes released by white blood cells in the lungs. If inherited, AAT deficiency can lead to emphysema.

Symptoms of AAT Deficiency (some or all)

- Shortness of breath
- Cough and/or sputum production
- Wheezing and recurring chest colds
- Jaundice
- Abdominal swellings and/or gastrointestinal bleeding
- Unexplained liver problems

Who should get tested?

All people with a diagnosis of chronic obstructive pulmonary disease (COPD), i.e. emphysema or chronic bronchitis, should be tested once for AAT deficiency. In addition, people with a diagnosis of asthma, whose breathing tests (performed by your doctor or at a pulmonary function laboratory) do not return to normal after inhaling a bronchodilator should be tested once in a lifetime for AAT deficiency. People with a family history of AAT deficiency, emphysema or liver disease at an early age, adult-onset asthma, or recurrent bronchitis should also be considered for testing. Testing is quick, easy, and highly accurate with a simple finger stick.

Treatment for AAT Deficiency

The highest priority for those with AAT deficiency is to quit smoking. As with non-hereditary COPD,

treatment should include scheduled vaccinations for influenza and pneumonia and compliance with prescribed inhaled medications to optimize lung function. Your physician may recommend a pulmonary rehabilitation program. Additionally, weekly intravenous infusions of alpha-1 antitrypsin may reduce symptoms, decrease the loss of lung function, and increase survival of patients. Lung transplantation may also be an option for those with severe disease.

Talking with your family

Since AAT deficiency is hereditary, people with this condition should talk with their family members and encourage them to be tested. Although others may not test positive for this disease, they may be carriers. Genetic and psychological counseling can provide knowledge and support for those families who have been affected by the disease.

Recommendations for those with AAT Deficiency

- Avoid all of the following: tobacco smoke, environmental and occupational pollutants, wood-burning stoves, fumes from household cleaning products, and paints and/or other toxic agents
- Maintain regular physician appointments and comply with prescribed medication and professional advice
- Stay current with immunizations
- Follow a good nutrition and exercise program
- Reduce alcohol consumption
- Develop a stress-management program
- Avoiding exposure to people who are sick
- Participate in Pulmonary Rehabilitation if recommended by your care provider