COPD and Alpha-1 Antitrypsin (AAT) Deficiency

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Overview

What is alpha-1 antitrypsin deficiency?

Alpha-1 antitrypsin (AAT) is a protein normally found in the lungs and the bloodstream. It helps protect the lungs from the damage caused by inflammation that can lead to emphysema and chronic obstructive pulmonary disease (COPD). People whose bodies do not produce enough of this protein (AAT deficiency) are more likely to develop emphysema and to do so at a younger-than-normal age (30 to 40 years old). AAT deficiency is a rare disorder and is the only known genetic (inherited) factor that increases your risk of developing COPD.

What puts you at risk for alpha-1 antitrypsin deficiency?

Your doctor may suspect you have an AAT deficiency if you:[1]

- Develop emphysema at 45 years of age or younger.
- Develop emphysema without having any recognized risk factors, such as smoking or inhaling industrial dust or chemical fumes over a long period of time.
- Develop emphysema, and X-rays show less density in the lungs than normal (basilar hyperlucency).
- Develop unexplained liver disease.
- Have a family history of emphysema, bronchiectasis, liver disease, or inflammation of the fat under the skin (necrotizing panniculitis).
- Have bronchiectasis without an evident cause.

Who should be tested?

An AAT deficiency test measures the level of AAT in the blood. The Global Initiative for Chronic Obstructive Lung Disease (GOLD) 2017 clinical guidelines recommend a one-time testing for all people with COPD.[2] And the American Thoracic Society and the European Respiratory Society recommend testing for an AAT deficiency for people who have:[1]

- Symptoms of COPD and have been diagnosed with emphysema or COPD.
- Asthma with airflow blockage that is not completely resolved after treatment with medicine that opens the lung airways (bronchodilators).
- Unexplained liver disease.
- No symptoms, but lung tests show obstruction, and risk factors such as smoking are present.
- Necrotizing panniculitis.
- A brother or sister who is AAT-deficient.

Screening for an AAT deficiency in the general public is not currently recommended.[1]

How is alpha-1 antitrypsin deficiency treated?

It is extremely important that you do not smoke if you have an AAT deficiency. Smokers with this condition may suffer devastating disease at a young age. People with this condition who have never smoked usually do not have significant symptoms at any age.

Treatment for COPD may include medicines to help you breathe easier. It may also include pulmonary rehabilitation. This means learning exercise, eating, and breathing tips and other ways to help yourself stay as healthy and strong as you can. And your doctor may suggest that you have injections of man-made alpha-1 antitrypsin protein (also called an alpha-1 proteinase inhibitor) that has been obtained from human plasma. Examples include Aralast, Prolastin, and Zemaira. To be considered for this treatment, you must meet the following guidelines:

- Your blood levels of the alpha-1 antitrypsin enzyme are less than 11 µmol/L (micromoles per liter).
- DNA testing shows that your body does not produce enough of the enzyme or produces an enzyme that does not work properly.
- You do not smoke or have stopped smoking.
- You have difficulty breathing because of COPD or emphysema.

Injections of replacement alpha-1 antitrypsin are given either weekly or every 2 to 4 weeks. Benefits of the therapy are not clear at this time.

Credits

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