



# medfacts

AN EDUCATIONAL HEALTH SERIES FROM NATIONAL JEWISH HEALTH™

## Alpha-1 Antitrypsin Deficiency or Inherited Emphysema

Chronic obstructive pulmonary disease or COPD for short, is a lung disease that affects millions of people each year. COPD is a general term used to describe diseases such as emphysema and chronic bronchitis. In emphysema there is damage to the walls of the air sacs (alveoli) in the lungs. The earliest symptom of emphysema is shortness of breath during activity. Later the shortness of breath can occur at rest also.

### What is inherited emphysema?

Most cases of emphysema are caused by smoking or other environmental factors. There is growing evidence that genetics can also increase the risk of developing emphysema. In about one out of every 50 cases of emphysema, there is a specific hereditary basis for the disease.

The inherited form of emphysema is called **Alpha-1 Antitrypsin Deficiency** or "Alpha - 1" for short. People with this disorder don't have enough of a major protein in the blood. This protein is alpha-1 antitrypsin. Although there are many different genetic variations of this protein, only some will cause lung disease. The most common variant of the protein is often seen in people of northern European descent, especially Scandinavians. Recent

evidence shows that Alpha-1 can be found in all populations and all ethnic groups.

### What does the alpha-1 protein do in the body?

Alpha-1 antitrypsin is a major protein in the blood. It is produced mainly in the liver cells. It protects the lung by blocking the effects of powerful enzymes called elastase. Elastase is normally carried in white blood cells and protects the delicate tissue of the lung by killing bacteria and neutralizing tiny particles inhaled into the lung. Alpha-1 antitrypsin inactivates elastase once it has finished its job. Without alpha<sub>1</sub> antitrypsin, elastase can destroy the air sacs of the lung.

### How is the diagnosis made?

Because inherited emphysema is a form of COPD, the diagnosis is made by the same methods. Your doctor may have you do a number of tests to evaluate your breathing. These may include:

- Detailed medical history including family history of lung disease
- Physical exam
- Breathing tests and X-rays
- Oxygen levels

Two special blood tests determine the diagnosis of inherited emphysema. The first test measures the concentration of alpha-1 antitrypsin in the blood. A second blood test determines the actual gene product in the person with the disease. Healthy people have an MM genetic pattern. This leads to normal levels of alpha-1 antitrypsin in the blood. The most common abnormal genetic pattern seen with inherited emphysema is ZZ. Some people may inherit only a single gene for inherited emphysema, such as MZ. It is unclear to what extent carrying a single abnormal gene for Alpha-1 increases the risk of emphysema.

## **How is inherited emphysema managed?**

Specific therapy has been available for inherited emphysema since 1987; a class of medicine called augmentation therapy. This medicine replaces the alpha-1 protein in the blood with normal alpha-1 antitrypsin from healthy plasma donors. It is given in a vein (IV). The dose is adjusted based on body weight. This treatment is often given once a week. There are three brands of augmentation therapy. They include:

- Prolastin®
- Aralast™
- Zemaira™

In addition to medicines, the management of inherited emphysema includes:

- Exercise and a healthy lifestyle
- Avoidance of infection
- Techniques to bring up mucus
- Oxygen therapy
- Pulmonary rehabilitation.

For more information, ask your health care provider for Med Facts on any of these topics.

Giving up smoking and avoiding secondhand smoke is very important. Smoking can accelerate or speed the development of the disease and shorten the lifespan.

Genetic counseling is important for family members of the person diagnosed with inherited emphysema. Family planning issues and early interventions, such as giving up smoking can be addressed.

Lung transplants or lung reduction surgery may be an option for people severely affected by the disorder.

It is important to appreciate that people with Alpha-1 may never develop emphysema. That is why Alpha-1 is referred to as a disorder or condition, rather than a disease. People with Alpha-1 are also at risk of developing liver disease and certain skin conditions. There may also be an association between Alpha-1 and atypical TB infections in the lung, also called non-tuberculous mycobacteria.

## **What does the future hold?**

Inherited emphysema is fairly common among inherited disorders. Research is constantly being done to develop new therapies. One promising therapy is replacement of the abnormal gene. Doctors at National Jewish Health follow one of the largest groups of Alpha-1 patients in the country. Virtually every new drug evaluated for this condition has been tested here. Doctors and researchers at National Jewish are always looking for new ways to manage and treat Alpha-1 and other chronic lung diseases.

Note: This information is provided as an educational service of National Jewish. It is not meant to be a substitute for consulting with your own physician.

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