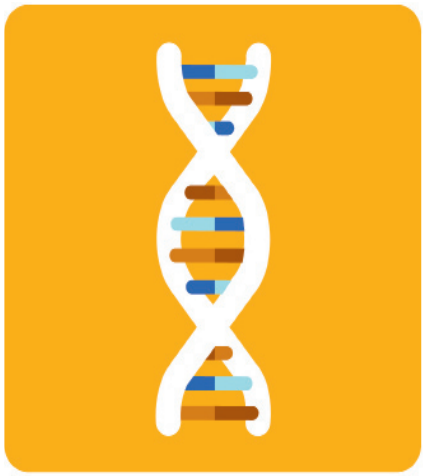
By Colleen Mullarkey

Alpha-$_1$ Testing and Treatment

While many consider chronic obstructive pulmonary disease a smoker's disease, the truth is it can strike non-smokers too. A hereditary condition called alpha-$_1$ antitrypsin deficiency, often called "genetic COPD," is the most commonly known genetic risk factor for the lung disease.

It occurs when there's a severe lack of alpha-$_1$ antitrypsin, a protein in the blood that protects the lungs from inflammation caused by infection and inhaled irritants. With early diagnosis and avoidance of risk factors like smoking, people with alpha-$_1$ may remain healthy throughout their lives.



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Who should be tested?

You should be tested for alpha-$_1$ if you have emphysema, COPD, chronic bronchitis, or asthma that's not completely reversible after aggressive treatment. Common respiratory symptoms include shortness of breath, wheezing, chronic cough, phlegm production, and recurring chest colds. If you have unexplained liver disease, a family history of liver disease, or a blood relative that has been diagnosed with alpha-$_1$, you should get checked out too.

Talk with your health care provider, who will need to give you a prescription for the test. Medical insurance usually covers the simple and highly effective blood test used to diagnose alpha-$_1$, but other resources also can provide free and confidential testing.

Your results will be analyzed at a laboratory, and generally you can expect an answer within one to two weeks. If the test shows you have inherited one defective alpha-$_1$ gene, you are an alpha-$_1$ carrier. It's unlikely that you will have any symptoms or require treatment, but you could pass the abnormal gene onto your children. If you have inherited two defective genes, you'll need to talk with your provider to determine the best treatment plan to manage your condition and symptoms.

Treatment options

You'll most likely visit a pulmonary specialist at least twice a year with additional follow-up based on your individual condition. The specialist usually will measure your oxygen levels with a simple, painless device called a pulse oximeter. A series of breathing tests called pulmonary function testing will determine how well your lungs are working. The doctor also may order lung X-rays and CT scans to take a closer look at your lungs' physical condition.

All of these tests will help to determine what kind of treatment you need. Smoking cigarettes is the biggest risk factor for lung disease and damage, so quitting is a must. In addition, your doctor may prescribe inhaled medications, antibiotics, or supplemental oxygen to treat your symptoms as needed.

Augmentation therapy, the standard treatment for alpha-$_1$-related lung disease is a weekly treatment that replaces the lacking protein through an IV. A professional can administer the treatment in your home, a clinic, or the hospital, or you can administer the treatment yourself. You should speak with your provider and your insurance company to determine the best option for you.

Receiving a diagnosis of alpha-$_1$ antitrypsin deficiency can be intimidating, but resources like the Alpha-1 Foundation can help by putting you in touch with health care professionals and other patients who understand your medical and emotional needs. n

Information provided by Bob Campbell, communications manager at the Alpha-1 Foundation and adapted from www.alphaone.org.

This Patient Primer was sponsored by:

ALPHA-1 FOUNDATION

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