

What Should I Do With the Results?

If your test results are positive:

- Contact your primary health care provider. Ask them about:
 - Interpretation of your test results
 - How the results will affect your health condition
 - The impact of the Alpha-1 on your current health state
 - Your options for treatment
- Stop smoking and avoid secondhand tobacco smoke as much as you can.
- Avoid being around dusts and fumes.
- Think about your health behavior (i.e. washing your hands, reducing contact with persons who have respiratory infections).
- Decide whom you should inform in your family and if they should think about being tested.
- Create an exercise program (under medical supervision).
- Create a nutrition program (under medical supervision).
- Ask your health care provider for a copy of the brochure "Guide to the Recently Diagnosed."
- Contact the resources listed below for more information.

There are ways that Alphas (persons with Alpha-1) can protect themselves through proper health care, eating well, exercise and stress management. Most importantly, Alphas can protect themselves by not smoking! In many cases, doing these things can help to fight off the symptoms of the disorder for many years.

Where Can I Go for More Information?

Alpha-1 Foundation

1-877-2-CURE-A1 (228-7321)
<http://www.alphaone.org>

AlphaNet

1-800-577-ANET (577-2638)
<http://www.alphanet.org>

Alpha-1 Association

1-800-521-3025
<http://www.alpha1.org>

American Liver Foundation

1-800-GO LIVER (465-4837)
<http://www.liverfoundation.org>

American Lung Association

1-800-LUNG-USA (586-4872)
<http://www.lungusa.org>



The Alpha-1 Foundation is a not-for-profit organization dedicated to providing the leadership and resources that will result in increased research, improved health, worldwide detection, and a cure for Alpha-1 Antitrypsin Deficiency (Alpha-1).

The Foundation provides the infrastructure to promote research and the development of new therapies for improving the quality of life for those diagnosed with Alpha-1. The Foundation is committed to close collaborations with medical experts, government agencies, international regulatory authorities, the pharmaceutical industry and other organizations to jointly resolve critical issues in the field of Alpha-1 research and treatment. A Grants Award Program supports a wide range of meritorious research in Alpha-1.

Telephone: 1-877-2-CURE-A1 (228-7321) Toll Free

Website: <http://www.alphaone.org>



The Alpha-1 Association is a nonprofit, membership organization founded in 1991. This grassroots social enterprise represents and is governed by the community of people who are affected by Alpha-1. Our mission is "to identify those affected by Alpha-1 Antitrypsin Deficiency and to improve the quality of their lives through support, education, advocacy and research." That mission is fulfilled through an international network of support groups, a Peer Guide program to help newly diagnosed individuals, and an array of educational materials. The Alpha-1 Association advocates for the community on a host of issues including genetic privacy and discrimination, insurance issues, and product safety and availability. The Association also encourages research and supports the programs of the Alpha-1 Foundation.

Telephone: 1-800-521-3025

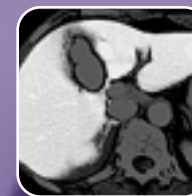
Website: <http://www.alpha1.org>

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What is Alpha-1?

Should I Be Tested?



What is Alpha-1?

Alpha-1, also known as Alpha-1 Antitrypsin Deficiency, is a disorder passed on by a person's parents, which results in low levels or no levels of a protein in the blood. This protein is called alpha₁-antitrypsin (AAT). If you have Alpha-1 you may get a severe lung and/or liver disease or pass the flawed AAT gene on to your children. In people lacking the AAT protein, the AAT made in the liver cannot be fully released into the blood.

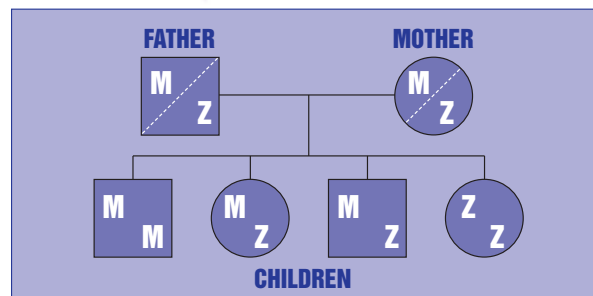
What Are Some Important Facts About Alpha-1?

Alpha-1:

- Is a disorder of the genes that leads to low or no levels of AAT
- May cause lung disease in adults
- May cause liver damage that gets worse overtime in adults, children and infants
- Is often not found or takes a long time to diagnose
- Can be treated, but cannot be cured
- Is easy to find through a blood test

How Is Alpha-1 Inherited?

One half of your genes are passed on from each parent. Refer to the figure below to see what could happen for children if both parents are carriers (have one normal and one altered AAT gene). Please note that this figure shows the most common normal and flawed genes.



Risks Associated with Common Genetic Variants

Normal (MM) Does not have the disorder and does not carry any altered AAT genes.

Carrier (MZ) Mild to moderate AAT Deficiency - may get disease symptoms and does carry an altered AAT gene.

Carrier (MS) It is unclear whether there is a risk for getting disease symptoms but does carry an altered AAT gene (most studies do not show an increased risk for disease).

Alpha-1 (SZ) or (ZZ) Moderate (SZ) to severe (ZZ) deficiency - could get disease and does carry two altered AAT genes.

Alpha-1 (SS) It is unclear whether there is a risk for getting disease symptoms but does carry two altered AAT genes (most studies do not show an increased risk for disease).

What Are The Signs Suggesting Alpha-1?

1. A Family History of: Alpha-1 or lung or liver disease

2. Symptoms

- Shortness of breath at rest or with exercise
- Wheezing
- Coughing
- Repeated lung infections
- Sputum (or phlegm) production
- History of suspected allergies and/or asthma

3. Any of these medical problems:

- Chronic Obstructive Pulmonary Disease (COPD)
- Emphysema
- Bronchiectasis
- Chronic bronchitis
- Asthma
- Chronic liver disease in adults
- The skin disease panniculitis
- Unexplained liver disease in infants and children

It is important to note that people with Alpha-1 may not show any signs of the disorder for many years. This does not mean that you will not have symptoms in the future.

What Is Involved In Testing For Alpha-1?

Testing for Alpha-1 is simple, quick and highly accurate. Your blood sample can be collected by means of a blood draw or by utilizing a finger stick method of blood collection.

Three types of tests may be conducted on your blood sample:

- Alpha-1 Antitrypsin Level test determines the amount of AAT in your blood
- Alpha-1 Antitrypsin PI Type or Phenotype test determines the type of AAT protein that you have
- Alpha-1 Genotyping examines your genes and determines your genotype

Testing Programs

Alpha-1 Screening and Detection Program

You or your health care provider may obtain an Alpha-1 Test Kit (finger stick) from the Alpha-1 Foundation, by calling toll free: (1-877-228-7321 ext. 217). Your health care provider will perform the test and submit the sample to the Alpha-1 Genetics Laboratory at the University of Florida. Results are returned to your health care provider within 2-4 weeks. This program gives your provider your AAT level and your AAT PI type (also called phenotype). Testing is performed free of charge through this program.

Alpha-1 Coded Testing (ACT) Trial

You may choose to be in a private testing program called the Alpha-1 Coded Testing (ACT) Trial. You may obtain an Alpha-1 Test Kit (finger stick) by calling the Medical University of South Carolina toll free at: (1-877-886-2383). You perform the test and the AAT genotype results are returned to you in 4-5 weeks. A counselor is available to discuss the results with you. There is no cost to you or your insurance for this testing service.

Commercial Laboratory

Your physician may decide to draw blood and send it to a commercial laboratory for analysis. The cost of the test varies and must be paid by you or your insurance company.

Who Should Consider Being Tested?

If you have signs that suggest Alpha-1 (family history of Alpha-1, specific symptoms or any of the identified medical problems) you should consider being tested.

There are ways your life could be affected by learning information that may be discovered by genetic testing.

Potential Benefits

- Allow you to increase your knowledge of Alpha-1 and awareness of your personal risk
- Provide information for future health care decisions
- Allow you to take steps that may slow the progression of Alpha-1
- Assist you and your family in making decisions about work, lifestyle and having children

Potential Harms

- May be personally unsettling
- May affect your ability to get health and life insurance
- May influence willingness of employers to hire you
- May create stress in your family
- May increase your personal health care costs

These tests are *up to you*. You should discuss health and non-health risks with your health care provider, family and others close to you. You need to understand the potential benefits and harms very well before testing. This is called informed consent.