ALPHA-1 FOUNDATION

WHAT IS ALPHA-1?



What you need to know about Alpha-1 Antitrypsin Deficiency (Alpha-1).



What is Alpha-1 Antitrypsin Deficiency?

Alpha-1 Antitrypsin Deficiency (Alpha-1) is a genetic condition that can cause damage to the lungs and/or the liver. It is a progressive condition, which means it may worsen over time. There is no cure for Alpha-1, but some symptoms can be managed with treatment. Alpha-1 occurs when there is a severe lack of a protein in the blood called alpha-1 antitrypsin (AAT) that is mainly produced by the liver. The main function of AAT is to protect the lungs from inflammation caused by infection and inhaled irritants such as tobacco smoke.

The low level of AAT in the blood occurs because the AAT is abnormal and cannot be released from the liver at the normal rate. This leads to a buildup of abnormal AAT in the liver that can cause liver disease.

What are some important facts about Alpha-1?

- Alpha-1 is a genetic condition that leads to low or undetectable levels of AAT
- Alpha-1 may cause lung disease in adults
- Alpha-1 may cause liver damage that gets worse over time in adults, children and infants
- Alpha-1 often goes undetected for years
- Alpha-1 can be treated, but cannot be cured without a liver transplant
- Alpha-1 is easy to detect through a simple blood test

Common Symptoms of Alpha-1

Symptoms related to the lungs:

- Shortness of breath, particularly with exertion
- Wheezing
- Chronic cough, often with sputum (phlegm) production
- Recurring chest colds
- Extreme tiredness (fatigue)





Symptoms related to the liver:

- Eyes and skin turning yellow (jaundice)
- Swelling of the abdomen or belly (ascites)
- Vomiting blood or passing blood in the stool
- Itchy skin



Risks associated with common genetic variants:

Normal (MM)
Does not have the condition; does not carry any abnormal AAT genes.

Carrier (MZ)
Mild to moderate Alpha-1 Antitrypsin Deficiency (Alpha-1)— may get disease symptoms and does carry an abnormal AAT gene.

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

Alpha-1 (SZ) or (ZZ)
Moderate (SZ) to severe (ZZ) Alpha-1 Antitrypsin Deficiency (Alpha-1) — could get disease and does carry two abnormal AAT genes.

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

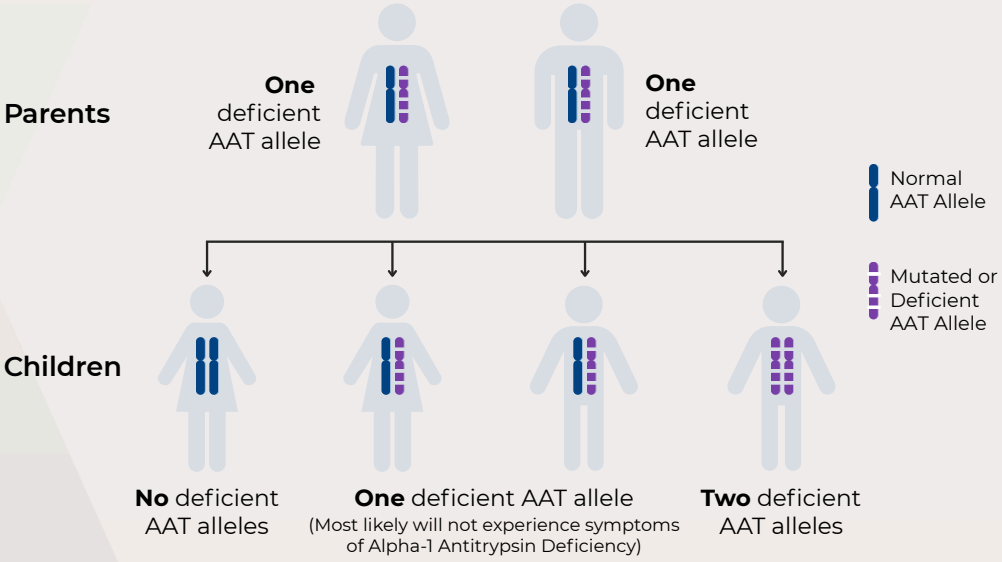




How is Alpha-1 inherited?

Half of your genes are passed on from each parent. You can inherit Alpha-1 from Alphas (people with two abnormal AAT genes) or carriers (people with one normal and one abnormal AAT gene).

Refer to the figure below to learn more about how both parents carry an abnormal gene. This figure shows the genes that result in normal AAT protein levels (M) and low to undetectable AAT protein levels (Z).



Testing & Diagnosis for Alpha-1

Why should people be tested for Alpha-1?

Many people with Alpha-1 are unaware they have the condition. Because Alpha-1 causes the same symptoms as more common conditions like COPD and asthma, it can prove especially challenging to diagnose. In fact, it can take several years for many people with Alpha-1 to be correctly diagnosed with the condition. If you suspect you or a family member has Alpha-1, talk to your doctor about getting tested.

Testing for Alpha-1 is the only way doctors can diagnose the condition. Diagnosing Alpha-1 early means getting treatment sooner, which may help slow or prevent lung damage and help you take better control of your health.

Because Alpha-1 is hereditary, getting tested is also important so Alpha-1 carriers can determine if they can pass the Alpha-1-causing gene on to family members. An Alpha-1 carrier is a person who has one normal Alpha-1 gene and one abnormal Alpha-1 gene. It should be considered when making decisions about having children, and should be discussed with your family members.

Testing for Alpha-1 is fairly simple, quick and highly accurate. It is done through a blood test or a mouth swab test.





Who should be tested for Alpha-1?

- People with COPD (chronic obstructive pulmonary disease), a group of lung diseases, including emphysema, and chronic bronchitis, that block airflow and make it difficult to breathe
- People with bronchiectasis
- Newborns, children and adults with unexplained liver disease
- People with panniculitis, a skin condition that some people with Alpha-1 develop
- Parents, siblings, children, and extended family members of people diagnosed with Alpha-1

Testing for Alpha-1

Alpha-1 can be diagnosed by a simple blood test. The test for Alpha-1 requires a physician's prescription and is usually covered by medical insurance. If the blood test shows lower than normal levels of AAT in the blood, the doctor might refer the person being tested for genetic counseling.

Though Alpha-1 is diagnosed by a blood test, other tests may be performed to determine the severity of a person's Alpha-1.

These tests may include:

- X-rays or CT scans of the lungs
- Pulmonary functioning tests (PFT)
- Liver ultrasound
- Liver biopsy (a small sample of tissue may be extracted)



Free Confidential Genetic Testing for Alpha-1

The Alpha-1 Foundation provides free, confidential genetic testing through the Alpha-1 Coded Testing (ACT) Study, in partnership with the University of Florida. The test can be done at home with a simple fingerstick. Results are usually received within 4-6 weeks. Scan the QR code below to request a test.

For more information, contact:

alpha1lab@alphaone.ufl.edu

(855) 476-1227

Before getting tested for Alpha-1

The Alpha-1 Foundation offers free, confidential genetic counseling services by phone to people with Alpha-1, family members of people with Alpha-1, and healthcare providers. These services are provided by the University of Florida. Our Genetic Counseling Services can provide:

- Information about testing, resources, prevention, and disease management
- Education about how Alpha-1 can be passed from parent to child
- Counseling to support families diagnosed with Alpha-1 and therapy referrals
- A review of available testing options for undiagnosed family members

To schedule a free appointment, call:

(855) 476-1227

Things to Consider When Testing for Alpha-1

Potential benefits of Alpha-1 testing:

- Deciding to stop cigarette smoking
- Choosing never to smoke
- Avoiding secondhand smoke
- Avoiding harmful exposures in your environment
- Avoiding excessive alcohol use
- Better conversations with healthcare providers about preventive care and improving health

Potential harms of Alpha-1 testing:

- May be personally unsettling
- May affect your ability to get life and disability insurance
- May create stress in your family
- May increase your personal healthcare costs

What should I do with the results of an Alpha-1 test?

- Contact your physician or primary healthcare provider
- Create an exercise & nutrition program (under medical supervision)
- Contact A1F for a copy of the “Guide for the Recently Diagnosed” brochure and view the resources listed on the last page of this brochure
- Decide whom to inform in your family and urge anyone who might be affected to get tested
- Think about your health behavior (smoking, alcohol use and excess weight) and avoid risk factors:
    - Stop smoking and avoid secondhand smoke
    - Avoid being around dust and fumes

Alpha-1 Foundation Programs, Services, & Resources

Alpha-1 Foundation (A1F)

alpha1.org | (877) 228-7321
The Alpha-1 Foundation provides resources, education, and information on testing and diagnosis for healthcare providers and people affected by Alpha-1. It funds cutting-edge research to find treatments and a cure and supports worldwide detection of Alpha-1.

A1F Patient Information Line

alpha1.org/patient-information-line | (877) 228-7321
Provides support to newly diagnosed Alphas and their families seeking basic information and help on a range of Alpha-1 related topics such as Alpha-1 testing, connecting with a peer guide, finding an Alpha-1 specialist, and requests for resources.

A1F Clinical Resource Centers (CRCs)

alpha1.org/find-an-alpha-1-specialist
An integrated network of research institutions and physicians specializing in Alpha-1 treatment, education, and care. CRCs provide comprehensive care to Alphas, including specialized care for lung disease and liver disease.

A1F Support Group Network

alpha1.org/find-a-support-group | (877) 228-7321
A collective of Alpha-1 support groups around the country committed to providing support and improving the quality of life of people affected by Alpha-1.



Alpha-1 Kids

alpha1.org/alpha-1-kids | (877) 228-7321
Information, support, and resources for families of Alpha infants, children, teens, and young adults transitioning to independent care.

The Alpha-1 Research Registry

alpha1.org/join-the-alpha-1-research-registry
(877) 228-7321 ext. 252
A confidential database of Alphas with one or two abnormal Alpha-1 genes that gives patients the opportunity to provide information to help advance research on the condition through questionnaires and clinical trials. It provides access to experts on Alpha-1 care. People enrolled have the ongoing opportunity to participate directly in clinical trials of new therapeutic approaches, in addition to other research opportunities.

A1F Genetic Counseling Services

alpha1.org/genetic-counseling | (855) 476-1227
A1F partners with the University of Florida to provide free genetic counseling services to Alphas, their family members, healthcare providers, and other individuals considering testing or in need of support. Genetic counseling services help patients understand their genetic risk for developing lung and liver disease, as well as communicating that information to family members.

ALPHA-1 FOUNDATION

The Alpha-1 Foundation (A1F) is committed to finding a cure for Alpha-1 Antitrypsin Deficiency (Alpha-1) and to improving the lives of people affected by Alpha-1 worldwide.

ALPHA1.ORG
1 (877) 2 CURE A1 | 1 (877) 228-7321

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