What is Alpha-1?

What you need to know about Alpha-1 Antitrypsin Deficiency

ALPHA-1 FOUNDATION

What Is Alpha-1?

Alpha-1 Antitrypsin Deficiency (Alpha-1) is a hereditary condition that is passed on from parents to their children through genes. This condition may result in serious lung disease in adults and/or liver disease in infants, children and adults.

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 the most common symptoms of Alpha-1?

Symptoms related to the lungs:

- Shortness of breath
- Wheezing
- Chronic cough and sputum (phlegm) production
- Recurring chest colds

Symptoms related to the liver:

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

What are some important facts about Alpha-1?

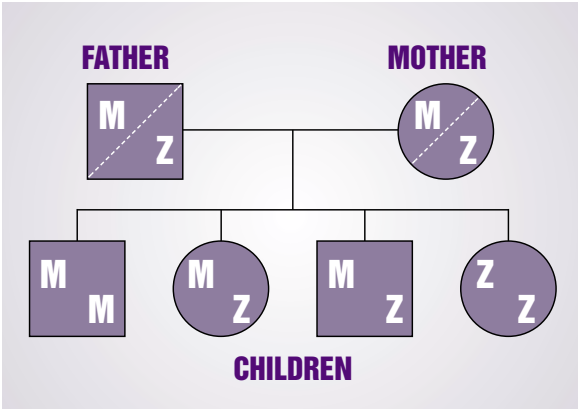
Alpha-1:

- Is a genetic disorder that leads to low or undetectable levels of AAT
- May cause lung disease in adults
- May cause liver damage that gets worse over time in adults, children and infants
- Often goes undetected for years
- Can be treated, but cannot be cured without a liver transplant
- Is easy to find through a blood test

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 see what could happen for children if both parents are carriers. This figure shows the genes that result in normal AAT protein levels (M) and low to undetectable AAT protein levels (Z).

Risks associated with common genetic variants



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

Carrier (MZ) • Mild to moderate AAT Deficiency — 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) deficiency — 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).

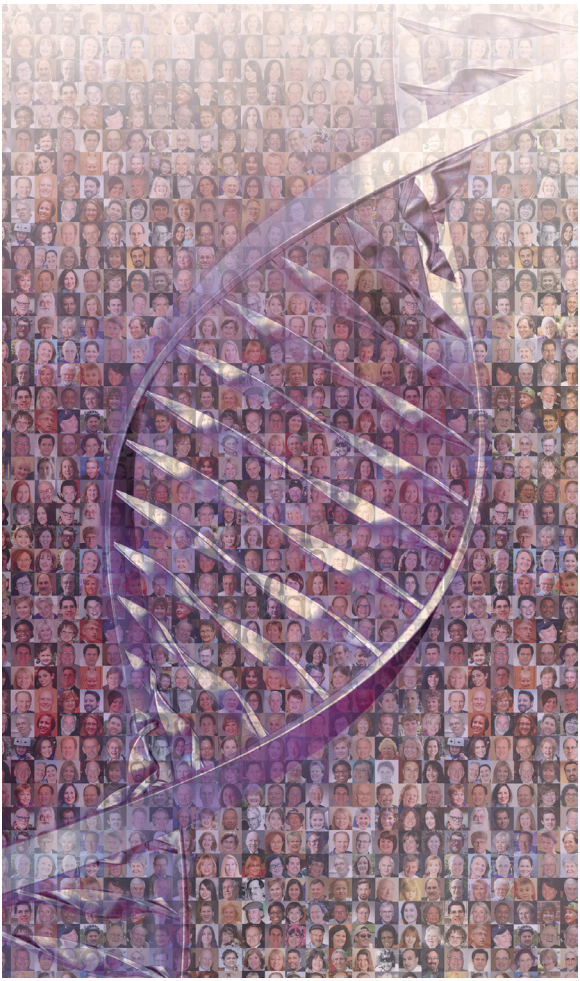
Who should be tested for Alpha-1?

- Everyone with emphysema, chronic obstructive pulmonary disease (COPD), or chronic bronchitis
- People with bronchiectasis
- Newborns, children and adults with unexplained liver disease
- People with a family history of liver disease
- Blood relatives of a person diagnosed with Alpha-1
- Anyone with panniculitis, a skin disease

Testing for Alpha-1

Testing for Alpha-1 is fairly simple, quick, and highly accurate. It is done through a blood test or a mouth swab test. People at risk for Alpha-1 should be tested because:

- There are treatments and preventive measures that may slow the progression of lung disease and help you take better control of your health.



- Since it is hereditary, Alpha-1 can be passed on to your children. It should be considered when making decisions about having children and should be discussed with your family members.

The test for Alpha-1 requires a physician's prescription and is usually covered by medical insurance. Confidential testing is available through the Alpha-1 Coded Testing (ACT) Study, through which anyone can have a free, confidential test and get their results. For more information on the ACT Study, call (877) 886-2383.

Potential benefits of Alpha-1 testing

- Allows you to increase your awareness of Alpha-1 and of your personal risk
- Provides information for future health care decisions
- Allows you to take steps that may slow the progression of Alpha-1
- Assists you and your family in making decisions about work and lifestyle
- Assists those of child-bearing age in understanding risk to children

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 health care costs

(For more information on Informed Consent, see *alpha1.org/testing-for-alpha-1*)

What should I do with the results?

- Contact your physician or primary healthcare provider.
- Create an exercise program (under medical supervision).
- Create a nutrition program (under medical supervision).
- Think about your health behavior (smoking, alcohol use and excess weight).
- Ask your healthcare provider for a copy of the brochure ***"Guide for the Recently Diagnosed."***
- Contact the resources listed in this brochure for more information.
- Avoid risk factors:
  - Stop smoking and avoid second-hand smoke as much as possible.
  - Avoid being around dust and fumes.
- Decide whom to inform in your family, and urge anyone who might be affected to get tested also.

RESOURCES

Alpha-1 Foundation
Toll Free: (877) 228-7321 • *www.alpha1.org*
The not-for-profit 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.

AlphaNet
Toll Free: (800) 577-2638 • *www.alphanet.org*
AlphaNet assists patients and families with support, education and strategies to manage their health. It also sponsors clinical trials for Alpha-1 therapies and produces ***The Big Fat Reference Guide to Alpha-1***, a complete guide to understanding, managing and living with Alpha-1, and includes key terms, testing, genetics, and treatment options. It is available through the website or number listed above.

The Alpha-1 Research Registry
Toll Free: (877) 886-2383
www.alphaoneregistry.org
The Research Registry is a confidential database of Alphas and carriers. The Registry gives patients the opportunity to provide information to help advance research on the disorder through questionnaires and clinical trials. It also provides access to experts on Alpha-1 care. People enrolled in the Registry have the ongoing opportunity to participate directly in clinical trials of new therapeutic approaches, in addition to other research opportunities.

Alpha-1 Foundation Patient Information Line
Toll Free: (800) 245-6809
The Patient Information Line is available free of charge to anyone affected by Alpha-1, and provides support to newly diagnosed Alphas, Alphas seeking basic information, or help such as requesting a peer guide, physician referral, requests for literature, etc.

American Association for the Study of Liver Diseases
(703) 299-9766 • *www.aasld.org*
This is the leading organization of scientists and healthcare professionals committed to preventing and curing liver disease.

Alpha-1 Foundation Genetic Counseling Program
Toll Free: (800) 785-3177
www.alpha1.org/genetic-counseling
The Genetic Counseling Program offers free, phone-based confidential information and resources to Alphas, family members and medical professionals. It provides information on the genetics of Alpha-1 and testing options.

Alpha-1 Foundation Support Network
Toll Free: (855) 351-6610
www.alpha1.org/support-groups
The Support Network is a collective of more than 80 Support Groups including four Virtual Support Groups. These groups are committed to improving the quality of life of people and their families affected by Alpha-1, and to extending the mission of the Alpha-1 Foundation.

Alpha-1 Kids
Toll Free: (877) 346-3212
www.alpha1.org/alpha-1-kids
Alpha-1 Kids provides support and information for parents and children with Alpha-1.

American Liver Foundation
Toll Free: (800) 465-4837
www.liverfoundation.org
The Foundation provides information on prevention, treatment and potential cures for liver diseases.

American Lung Association (ALA)
Toll Free: (800) 548-8252 • *www.lung.org*
The ALA focuses on the prevention of lung disease through educational programs, research and advocacy.

Children's Liver Association for Support Services
Toll Free: (877) 679-8256 • *www.classkids.org*
This group serves the emotional, educational and financial needs of families and children with liver disease.

Childhood Liver Disease Research Network
www.childrennetwork.org
The Consortium provides support and information for children and families with rare, cholestatic liver diseases. For information on liver issues related to AAT, visit this website and click on the Alpha-1 link.

ALPHA-1 FOUNDATION

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

www.alpha1.org
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Unrestricted educational grants provided by:

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