

Am I an Alpha-1 Carrier?



Learn how being an
Alpha-1 carrier can affect
you and your family



ALPHA-1
FOUNDATION

What Is Alpha-1 Antitrypsin Deficiency?

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 or adults.

For the most part, for each trait a person has there are two genes. One gene comes from each parent. People with Alpha-1 have received two defective alpha-1 genes. One defective gene came from their mother and one from their father. There are many types of defective alpha-1 genes. The most common of these are called S or Z. Normal genes are called M. A person who does not have Alpha-1 will have two M genes (MM). People with Alpha-1 most commonly have two Z genes (ZZ). About 20% of people with SZ genes will have lung and/or liver problems like those of people with ZZ genes.

The result of having two defective alpha-1 genes is a very low or even absent level of a protein called alpha-1 antitrypsin (AAT) in the blood. The deficiency in the blood is caused by an abnormal accumulation or absence of the alpha-1 protein in the liver.

What is meant by the term “Alpha-1 carrier”?

An Alpha-1 carrier is a person who has one normal alpha-1 gene (M) and one defective alpha-1 gene (usually Z or S). Being a carrier is very common. It is believed that over 19 million people in the United States are carriers. Most Alpha-1 carriers are MZ or MS. Carriers may have lower blood levels of the alpha-1 antitrypsin protein, but their levels are rarely as low as those of people with Alpha-1.

How can being an Alpha-1 carrier affect your lungs?

Alpha-1 carriers usually have only a slight risk of developing a disease related to Alpha-1. The main type of carrier linked to increased risk for lung diseases has MZ genes. Currently, there is no known risk for lung diseases for MS carriers.



A special thanks from the Alpha-1 Foundation to Vermont Alpha Vicki Cameron and her family who allowed us to use their photo. Vicki is second from the left.

Lung Disease: The risk for emphysema may be greater for MZ carriers. This increased risk, however, is very small unless the carrier is a smoker or exposed to high levels of air pollution. The risk of having Chronic Obstructive Pulmonary Disease (COPD) is higher among MZ carriers who have relatives with COPD. This suggests that the COPD in these families may be due to other genetic factors.

Lung symptoms that might be linked to being an Alpha-1 carrier:

- Shortness of breath
- Wheezing
- Chronic cough and sputum (phlegm) production
- Chronic bronchitis
- Recurring chest colds
- Decreased exercise tolerance
- Year-round allergies
- Bronchiectasis

How can being an Alpha-1 carrier affect your liver?

Alpha-1 carriers usually have only a slight risk of developing a disease related to Alpha-1. The main type of carrier linked to increased risk for liver diseases has MZ genes. Currently, there is no known risk for liver diseases for MS carriers.

Liver Disease: The risk of chronic liver disease in Alpha-1 carriers is much less than that for people with Alpha-1. Research suggests that chronic liver disease might appear in MZ carriers only when the liver has been damaged first by something else, such as a virus, chemicals including alcohol or being overweight.

Liver symptoms that may be related to carrier status:

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

Children of Alpha-1 carriers

Alpha-1 carriers may pass their defective alpha-1 gene to their children.

- If a carrier (MZ) has a child or children with a person who has normal alpha-1 genes (MM), each child has one chance in two (50% risk) of being an Alpha-1 carrier (MZ). There is no risk that any of the children will have the condition.
- If a carrier (MZ) has children with another carrier (MZ), each child has one chance in two (50% risk) of being an Alpha-1 carrier. Each child also has one chance in four (25% risk) of having Alpha-1 (ZZ) and one chance in four (25%) of having normal alpha-1 genes (MM).



Who should be tested?

Anyone thinking about being tested for Alpha-1 should speak first with a healthcare professional that has knowledge of genetic diseases. This could be their physician or a genetic counselor. Testing is advised for parents, brothers, sisters and adult children of a person with Alpha-1.

Testing is also advised for anyone with the following medical conditions:

- COPD (emphysema and/or chronic bronchitis)
- Unexplained liver disease
- Liver disease with a family history of liver disease

If you are a carrier and are considering having children, see www.alpha1.org for more detailed information.



How can I be tested?

Finding out your Alpha-1 status usually involves blood tests, which will indicate whether you are classified as normal (no Alpha-1 deficiency), carrier or deficient. However, tests of cells scraped from the inside of the cheek are also used. The most common test measures the amount of alpha-1 antitrypsin protein in the blood. Many clinical laboratories can run this test. A second type of blood test or set of tests are called phenotyping and genotyping. Alpha-1 testing is mainly done only when the amount of AAT in the blood is below normal. These tests pinpoint exactly what type of alpha-1 antitrypsin protein the body makes. Only a few specialized labs can run these tests. There is a free confidential testing program called the Alpha-1 Coded Testing (ACT) Study at the Medical University of South Carolina (MUSC). To have family members tested to determine their status, please call (877) 886-2383 or email inquiries to alphaone@musc.edu.

Are there risks to being tested for Alpha-1?

Yes. Though the Genetic Information Nondiscrimination Act (GINA) provides some protections against discrimination regarding health insurance and employment, other forms of insurance such as disability are not covered by GINA. Once a test result is in your health record, third parties may get the information. Third parties may include insurance companies, healthcare centers or other professionals. They may learn your test result if you sign a release form that gives access to your records.

Informed consent

Informed consent is the process through which a person receives appropriate information, understands that information and agrees to testing. It originates from the legal and ethical right the patient has to direct what happens to their body and from the ethical duty of the physician to involve the patient in their healthcare. You should discuss the decision to get tested for Alpha-1 with your doctor and make sure all of your questions are answered. (For more information on informed consent, please go to www.alpha1.org.)

Can being a carrier affect my health insurance?

Generally, no. It is against the law for your health insurer to use a genetic test result or family health history as a reason to deny you health insurance, or decide how much you pay for your health insurance. Insurers may not request a genetic test, or use genetic information to discriminate against you.

How can carriers prevent or reduce their risk of getting the diseases linked to Alpha-1?

You may prevent or reduce the risks by making changes to your lifestyle, such as:

- Do not smoke and avoid second-hand smoke.
- Avoid repeated exposure to dust, fumes or gases.
- Quit or cut back on drinking alcohol.
- Get vaccinated against hepatitis A and B.

If the carrier has children who are also carriers, the children should be informed about their genetic status. The importance of a healthy lifestyle should be emphasized from an early age.

What are the recommended treatments for carriers with liver or lung disease?

Your doctor will determine the course of your treatment. Treatment to correct the decreased blood levels of AAT in Alpha-1 carriers is not recommended. AAT augmentation therapy, which is costly, is only indicated for those with emphysema, who have a severe deficiency of AAT.

To whom should I reveal my Alpha-1 carrier status?

Sharing your status with others is up to you, but you may want to inform family members so they can consider testing. Also, your physician may need to know to better plan treatment options.

Where can I go for more information and support?

Learning that you are an Alpha-1 carrier may confuse or upset you. It may help you to:

- Share your status with family.
- Learn as much as you can about the effects it can have on your health.
- Seek support groups or genetic counseling to answer your questions.

The Alpha-1 Patient Information Line can answer many of your questions. Call 1-800-245-6809.



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. Individuals 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

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

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 individuals 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/support/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.



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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