



Research for a Cure

The mission of the **Alpha-1 Foundation** is to provide the leadership and resources that will result in increased research, improved health, worldwide detection, and a cure for Alpha-1 Antitrypsin Deficiency. The Foundation has invested nearly \$39 million to support Alpha-1 Antitrypsin Deficiency research in more than 70 insitutions in North America, Europe and Australia. For more information, visit: www.alpha-1foundation.org.



Support, Education and Advocacy

The **Alpha-1 Association** is the leading national patient membership organization serving the Alpha-1 community since 1991. Its mission is to identify those affected by Alpha-1 Antitrypsin Deficiency and to improve the quality of their lives through support, education, advocacy and to encourage participation in research. Its programs include a national Support Network of more than 70 Support Groups, Patient Hotline, national and regional educational programs, Genetic Counseling Service, and grassroots advocacy program. For more information visit: www.alpha1.org.

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Unrestricted educational grants provided by:

Accredo Therapeutics
AlphaNet
Centric Health Resources
CSL Behring
Talecris Biotherapeutics



What Does It Mean to be an Alpha-1 Carrier?



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). It is rare to have both an S gene and a Z gene (SZ). 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 S or Z). Being a carrier is very common. It is believed that over 20 million people in the United States are carriers. Most Alpha-1 Carriers are MS or MZ. Carriers may have lower blood levels of 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.

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. There is no scientific evidence that MS carriers are at risk for lung disease.

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
- Non-responsive asthma or 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. There is no scientific evidence that MS carriers are at risk for liver disease.

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 and sisters of a person with Alpha-1.

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

- COPD (emphysema and/or chronic bronchitis)
- Asthma where lung function is not made normal by prescribed medications
- Unexplained liver disease
- Liver disease with a family history of liver disease

If you are a carrier and are considering having children, please go to www.alpha-1foundation.org for more information.

How can I find out the status for myself or someone I care about?

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 starting to be 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@muscu.edu

Is there any downside to being tested for Alpha-1

Yes. Genetic discrimination has occurred in the United States in employment and health and life insurance. 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.alpha-1foundation.org.)

Can being a carrier affect my health insurance?

The answer to this question is generally “No,” but in the future, insurance companies may view the carrier state as a pre-existing condition.

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

MZ carriers have only a slightly increased risk for the lung or liver diseases seen in people with 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 replacement 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.

Also, there are organizations that can offer help and advice to you. Some of these are listed in this brochure.



RESOURCES

Alpha-1 Association

Toll Free: (800) 521-3025

www.alpha1.org

The Association is a member-based nonprofit organization helping to identify those affected by Alpha-1, and dedicated to improving the quality of their lives through support, education and advocacy. It has a network of more than 70 volunteer-led support groups and a toll-free confidential Genetic Counseling Program that serves as an expert resource for patients, family members and health professionals. The certified genetic counselor is available at 1-800-785-3177.

Alpha-1 Foundation

Toll Free: (877) 228-7321

www.alpha-1foundation.org

The not-for-profit Foundation provides resources, educational brochures and information on testing and diagnosis for physicians and patients. 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 Kids

1 (877) 346-3212

www.alpha1kids.org

Alpha-1 Kids provides support and information for parents and children with Alpha-1.

The American Association for The Study of Liver Disease

(703) 299-9766

www.aasld.org

This is the leading organization of scientists and healthcare professionals committed to preventing and curing liver disease.

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: 1 (800) 548-8252

www.lungusa.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.

Cholestatic Liver Disease Consortium

(303) 837-2598

www.childrennetwork.org/families.html

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.



*A special thanks from the Alpha-1 Foundation to Vermont Alpha **Vicki Cameron and her family**. They allowed us to use their photo on the cover of this brochure. Vicki is second from the left on our cover.*