




# Alpha-1 Antitrypsin Deficiency: It's All in the Family





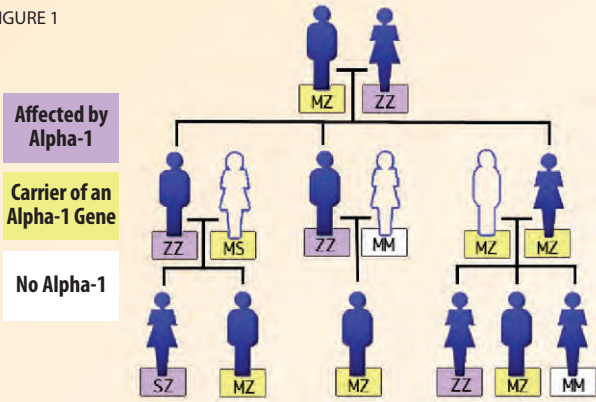
Whether you have Alpha-1 Antitrypsin Deficiency (Alpha-1) — or are an Alpha-1 “carrier” — we strongly suggest that your relatives be tested. Doing so could improve their lives and health.

Please give this brochure to your relatives and share with them what you have learned about Alpha-1.

# How is Alpha-1 inherited?

Your genes are inherited from your parents. If you have Alpha-1, you received one Alpha-1 gene from your mother and one Alpha-1 gene from your father. Your siblings may have inherited the same genes, or a different set of Alpha-1 genes. Most people identified with Alpha-1 have the ZZ "genotype". Another common gene for Alpha-1 is the "S" gene. Figure 1 shows an example of how Alpha-1 can be inherited.

FIGURE 1



*\*Spouses are indicated by a blue outline*

## Why should relatives be tested?

Your relatives may also have one or two Alpha-1 genes and not be aware of their own health risks. If they learn they have Alpha-1, they may consider different lifestyles, professions or other personal decisions that could maintain or improve their health. Their doctor may also be able to give them better medical advice. The benefits of testing include:

- Deciding to stop cigarette smoking, seeking help if necessary;
- Choosing never to smoke;
- Avoiding secondhand smoke;
- Avoiding harmful environmental and occupational exposures
- Avoiding excessive alcohol use
- A better dialogue with healthcare providers about preventive care and improving health.

## **Is there harm in knowing the results of family testing?**

There are laws protecting against some discrimination on the basis of genetic risk. The Alpha-1 Foundation has been encouraged by the passage of the Genetic Information Nondiscrimination Act (GINA) to move forward with a family awareness program. However, it is still unclear what protections are in place for those with Alpha-1. The Foundation recommends using the Alpha-1 Coded Testing (ACT) Study for family testing (page 4).

Knowing the results of genetic testing may create emotional stress. Genetic counselors can help with understanding and coping with these issues.

See the Foundation website, [www.alpha-1foundation.org](http://www.alpha-1foundation.org) for more on this subject. On the "Alphas, Friends & Family" page, click on the link "Get Tested" and read the section on "Informed Consent". Or call the Foundation at 1-800-425-7421.

## **What goes into the medical record?**

The results of every laboratory test ordered by your healthcare provider will be in your medical record. However, testing is available that is not entered into the medical record unless requested by the patient. See "How can a family member be tested?" on page 4.



## **Which relatives are at greater risk of Alpha-1?**

If you have Alpha-1, your immediate relatives — your children, parents, brothers and sisters — are at greater risk of having the S or Z genes. Other relatives who have lung or liver disease are also considered at greater risk.

## **Is Alpha-1 related only to the S and Z genes?**

There are rarer forms of Alpha-1 deficiency genes in addition to the S and Z genes. You should discuss family testing with a healthcare provider knowledgeable about Alpha-1, such as the Clinical Resource Centers on the Foundation website. On the “Alphas, Friends & Family” page, click on the link “Clinical Resource Centers”.



## **Should young children be tested?**

Currently, there is no newborn screening program for Alpha-1 in the United States. Based on the discussion above regarding genetic discrimination, we do not currently recommend testing children unless the child has symptoms of liver disease. We recommend testing both parents first. You may then wish to discuss this decision with your personal physician, your child's pediatrician, and a genetic counselor familiar with Alpha-1. It is important to discuss testing with your child as well, if your child is old enough to participate in the decision.

## **Can any testing predict whether my children will inherit Alpha-1 genes?**

A physician can make an exact prediction only if each parent has two of the same Alpha-1 genes (for example, MM in the father and ZZ in the mother). In this case, each child would have the MZ combination — an M gene from the father and Z gene from the mother. A healthcare provider can describe the possible genetic patterns that a child could have, only by knowing the exact genetic patterns of the child's biological parents. We recommend discussing this issue with a genetic counselor.

## **How can a family member be tested?**

For testing relatives, we recommend a genetic test, not a test of the alpha-1 antitrypsin level in blood. Options for the genetic test include:

- **Alpha-1 Coded Testing (ACT Study)** – No cost and confidential results are returned directly to the patient
  - Ask for a test kit from the Medical University of South Carolina. Call 1-877-886-2383 or email [alphaone@musc.edu](mailto:alphaone@musc.edu).
  - Answer a questionnaire and perform the test at home with a finger stick. Blood drops must cover three small circles on a card. This card gets sent back for testing.
  - Results are returned to the individual 4-6 weeks later.

- A certified genetic counselor familiar with Alpha-1 is available to discuss results. Call toll-free 1-800-785-3177.
- **Commercial laboratory** – A healthcare provider orders a blood test called alpha-1 antitrypsin genotyping. This is paid for by the patient or through healthcare insurance. The results are sent to the physician and are entered into the medical record.

## **How is a genetic counselor helpful?**

Genetic counselors are healthcare professionals who specialize in providing information about inherited conditions. A genetic counselor can help someone with the decision on whether or not to be tested. A genetic counselor can also help someone understand the test results and explain the results to their family. We recommend calling the Alpha-1 Association Genetic Counseling Program at 1-800-785-3177. This is a free and confidential service. You may also ask your doctor for a referral to a genetic counselor familiar with Alpha-1.

Please tell your relatives about Alpha-1 and encourage them to be tested. This will help to identify the many undiagnosed people with Alpha-1. Understanding one's genetic risk for liver and lung disease may result in an improved life and more informed health choices.

## **Be tested for Alpha 1: It's all in the family.**

For more information, call the Alpha-1 Foundation at 1-800-425-7421 or email [info@alphaone.org](mailto:info@alphaone.org).



Please fill out this brief letter. Then tear the page off this brochure and give to your relative to encourage Alpha-1 testing.

### **A letter to my**

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*Dear .....,*

*I want to share this brochure about Alpha-1 Antitrypsin Deficiency (Alpha-1) with you. I have inherited the genes for Alpha-1, which puts me at risk for lung and liver disease.*

*Since Alpha-1 runs in families, each member of our family should consider getting tested. The test results can help you to make good choices that promote health.*

*I would be happy to discuss this brochure with you.*

*Sincerely,*

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### 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 \$35 million to support Alpha-1 Antitrypsin Deficiency research in nearly 70 institutions in North America, Europe and Australia. For more information, visit: [www.alpha-1foundation.org](http://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 75 Support Groups, Patient Hotline, national and regional educational programs, Genetic Counseling Service, and grassroots advocacy program. For more information visit: [www.alpha1.org](http://www.alpha1.org).

[www.alpha-1foundation.org](http://www.alpha-1foundation.org)  
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