



## ALPHA-1: IT'S ALL IN THE FAMILY



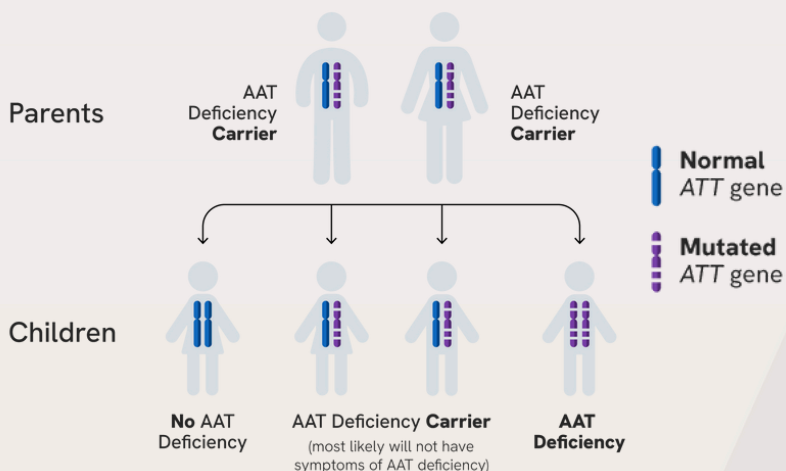
Testing for Alpha-1 Antitrypsin  
Deficiency (Alpha-1) could  
improve the lives and health  
of your relatives



If you have Alpha-1 Antitrypsin Deficiency (Alpha-1), it is strongly suggested 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. Since each individual has two genes (or alleles) for Alpha-1, your siblings may inherit the same genes as you, 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. The “M” gene is normal. Figure 1 shows an example of how Alpha-1 can be inherited.



## Why should relatives be tested?

Relatives of people diagnosed with Alpha-1 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 or carry a single abnormal Alpha-1 gene, 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 (A1F) 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. A1F recommends family testing (go to page 6 for more information).

Knowing the results of genetic testing may create emotional stress. Genetic counselors can help with understanding and coping with these issues. See page 7 for more information about genetic counseling.

Visit [alpha1.org/about-alpha-1-testing-diagnosis/](http://alpha1.org/about-alpha-1-testing-diagnosis/) for more on this subject.

## What goes into your 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 6.



## Which relatives are at greater risk for 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 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 Alpha-1 Specialists / Clinical Resource Centers (CRCs) on the A1F website. More information is available at [alpha1.org/find-an-alpha-1-specialist/](http://alpha1.org/find-an-alpha-1-specialist/).



# Children and Testing for Alpha-1

## Should young children be tested for Alpha-1?

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/or 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. See page 7 for more information about genetic counseling.





# Testing for Alpha-1

## How can a family member be tested for Alpha-1?

For testing relatives, we recommend a genetic Alpha-1 test, not a test of the Alpha-1 Antitrypsin (AAT) level in the blood.


## Free Confidential Genetic Testing for Alpha-1

The Alpha-1 Foundation (A1F) 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](mailto:alpha1lab@alphaone.ufl.edu)

 (855) 476-1227

Many people find it helpful to talk to a nurse who understands genetics and can provide counseling services and discuss test results.

You also have the option to test through a 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.

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

For more information, call A1F at 1-877-228-7321, ext. 326 or email [info@alpha1.org](mailto:info@alpha1.org)




## 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.

## Alpha-1 Foundation Genetic Counseling Program

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.

**To schedule a free appointment, call:**

 [\(855\) 476-1227](tel:(855)476-1227)

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.

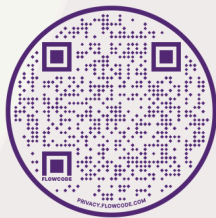
# Alpha-1 Foundation Programs, Services, & Resources



## Alpha-1 Foundation (A1F)

[alpha1.org](http://alpha1.org) | (877) 228-7321

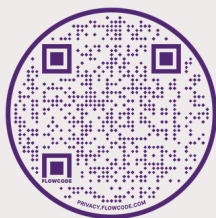
The Alpha-1 Foundation provides resources, education, and information on testing and diagnosis for healthcare providers and people affected by Alpha-1. It funds cutting-edge research to find treatments and a cure and supports worldwide detection of Alpha-1.



## A1F Patient Information Line

[alpha1.org/patient-information-line](http://alpha1.org/patient-information-line) | (800) 245-6809

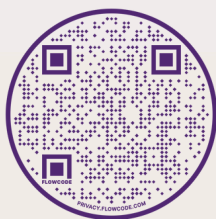
Provides support to newly diagnosed Alphas and their families seeking basic information and help on a range of Alpha-1 related topics such as Alpha-1 testing, connecting with a peer guide, finding an Alpha-1 specialist, and requests for resources.



## A1F Clinical Resource Centers (CRCs)

[alpha1.org/find-an-alpha-1-specialist](http://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.

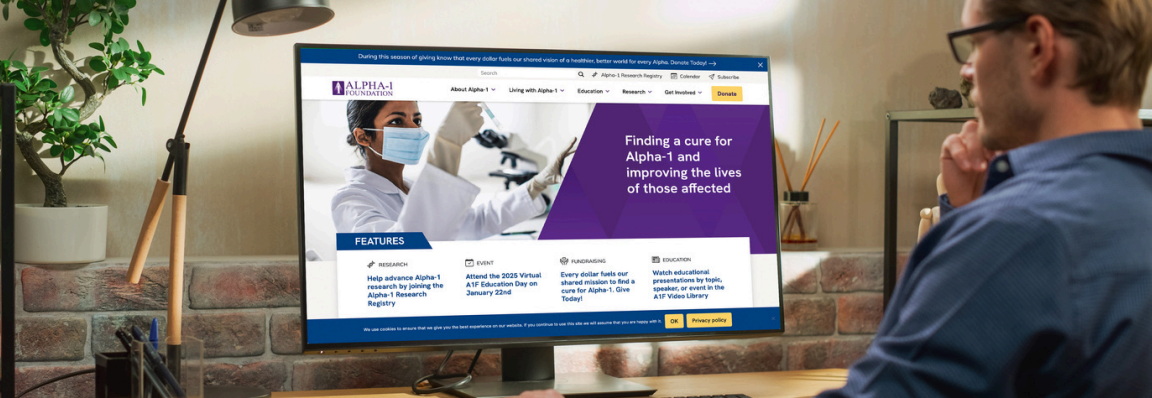


## A1F Support Group Network

[alpha1.org/find-a-support-group](http://alpha1.org/find-a-support-group) | (877) 346-3212

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](https://alpha1.org/alpha-1-kids) | (877) 346-3212  
Information, support, and resources for families of Alpha infants, children, teens, and young adults transitioning to independent care.



## The Alpha-1 Research Registry

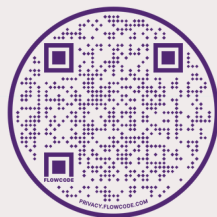
[alpha1.org/join-the-alpha-1-research-registry](https://alpha1.org/join-the-alpha-1-research-registry)  
(877) 228-7321 ext. 252

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](https://alpha1.org/genetic-counseling) | (855) 476-1227  
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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

Unrestricted educational grants  
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