



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.

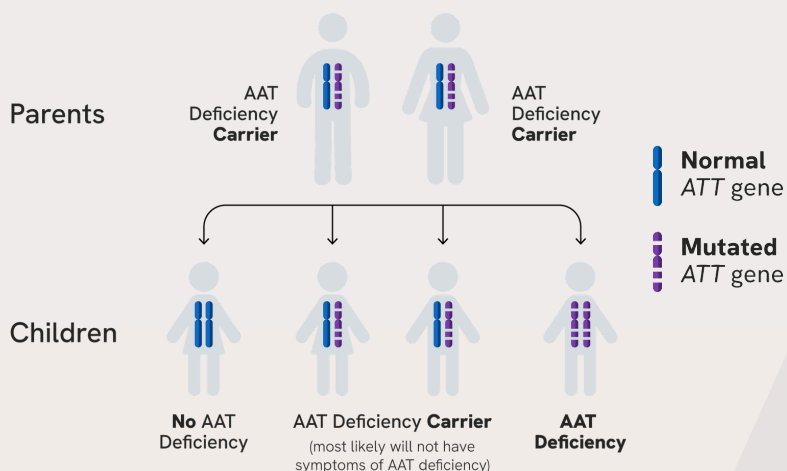


If you have Alpha-1 Antitrypsin Deficiency (Alpha-1), it is strongly suggested that your relatives be tested. Doing so could improve their lives and health. Please give this brochure to your relatives and share with them what you have learned about Alpha-1.

What Causes Alpha-1?

Alpha-1 is passed down from parents to children through genes. People with Alpha-1 have mutations in a gene called SERPINA1. The SERPINA1 gene provides instructions for making a protein called alpha-1 antitrypsin (AAT), which protects the body from damage by its immune cells.

If you have Alpha-1, you inherited two copies of the abnormal SERPINA1 gene—one from each parent.



What is an Alpha-1 carrier?

People who inherit one defective copy of the SERPINA1 gene and one normal copy of the SERPINA1 gene are known as Alpha-1 carriers. An estimated 19 million people in the United States are Alpha-1 carriers.

Alpha-1 carriers can pass the abnormal SERPINA1 gene to their children. If both parents are Alpha-1 carriers, their children have a 1 in 4 chance of inheriting Alpha-1 and a 1 in 2 chance of being an Alpha-1 carrier.

More about Alpha-1 Mutations

More than 150 different mutations in the SERPINA1 gene have been identified, but only a few can cause serious lung, liver or skin disorders. The most common of these are called S and Z:

- **ZZ:** People who inherit two ZZ mutations—one from each parent—have extremely low levels of AAT in the blood, which increases their risk of developing lung disease.
- **SZ:** People who inherit an S mutation and a Z mutation—one from each parent—have very low levels of AAT in the blood, which increases their risk of developing lung disease. However, people with SZ mutations are less likely to develop lung disease than people with ZZ mutations.

Many people with Alpha-1, including those with SZ or ZZ mutations, have no symptoms of the condition.

Testing for Alpha-1

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

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.

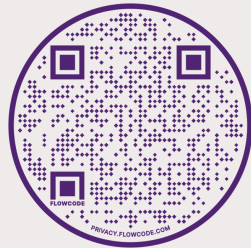
Testing for Alpha-1 (cont'd)

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/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/.



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.



Alpha-1 Testing Support

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



(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. See page 9 to learn more about genetic counseling.

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 about testing for Alpha-1, call A1F at 1-877-228-7321 or email info@alpha1.org.



Alpha-1 Foundation Genetic Counseling Services

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)

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.



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 lead to improved quality of life and more informed health choices.

Alpha-1 Foundation Programs, Services, & Resources



Alpha-1 Foundation (A1F)

alpha1.org | (877) 228-7321

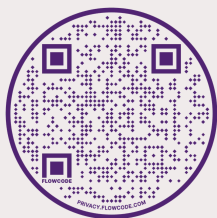
The Alpha-1 Foundation provides resources, education, and information on testing and diagnosis for healthcare providers and people affected by Alpha-1. It funds cutting-edge research to find treatments and a cure and supports worldwide detection of Alpha-1.



A1F Patient Information Line

alpha1.org/patient-information-line | (877) 228-7321

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

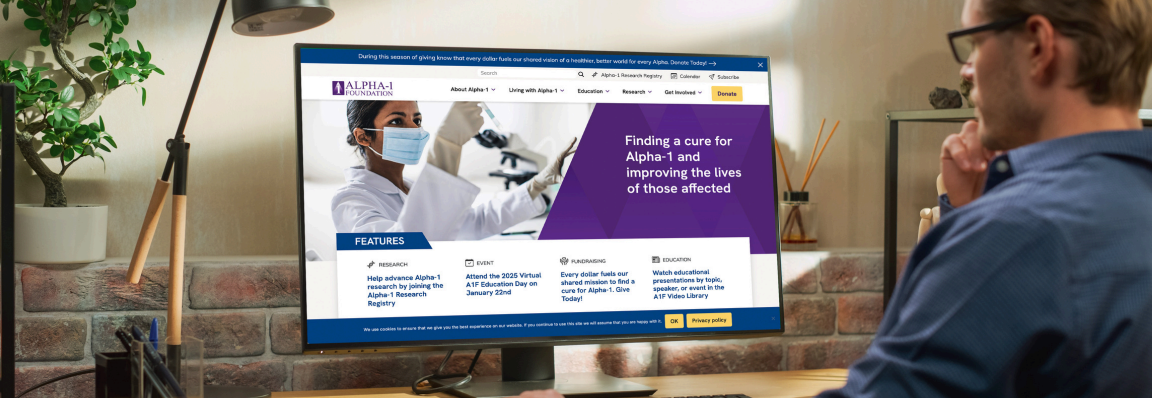
An integrated network of research institutions and physicians specializing in Alpha-1 treatment, education, and care. CRCs provide comprehensive care to Alphas, including specialized care for lung disease and liver disease.



A1F Support Group Network

alpha1.org/find-a-support-group | (877) 228-7321

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

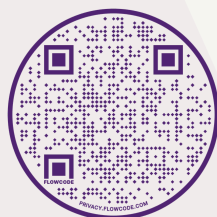
alpha1.org/alpha-1-kids | (877) 228-7321
Information, support, and resources for families of Alpha infants, children, teens, and young adults transitioning to independent care.



The Alpha-1 Research Registry

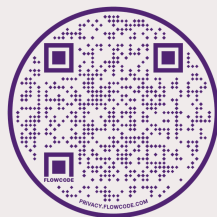
alpha1.org/join-the-alpha-1-research-registry
(877) 228-7321 ext. 252

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 | (855) 476-1227
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.





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

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