

## Genetic Counseling Program

The Alpha-1 Foundation Genetics Program offers experience in medical genetics and counseling. The program augments your care, as the healthcare professional, by addressing the social impacts of Alpha-1 with patients, families, and caregivers, and by providing you with advice and counsel regarding how to discuss the effect Alpha-1 may have on your patients.

The Genetics Counseling program provides an additional resource for the healthcare professional. The Genetics Counseling program provides expertise that may help you as a healthcare professional understand more about the genetic nature of Alpha-1, and the psychosocial and economic impacts on family members. The Genetics Counseling program can also give you specific guidance on the risks and benefits of genetic testing.

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For more information about the Alpha-1 Foundation, visit our website at [www.alpha1.org](http://www.alpha1.org) or call 1-877-228-7321.

The Alpha-1 Foundation Genetic Counseling Program is conducted by the University of Florida.



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.

The Foundation also offers a wide array of programs for people diagnosed with Alpha-1, their families, caregivers, and healthcare providers.

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[www.alpha1.org](http://www.alpha1.org)  
1 (877) 2 CURE A1 | 1 (877) 228-7321  
3300 Ponce de Leon Blvd.  
Coral Gables, FL 33134

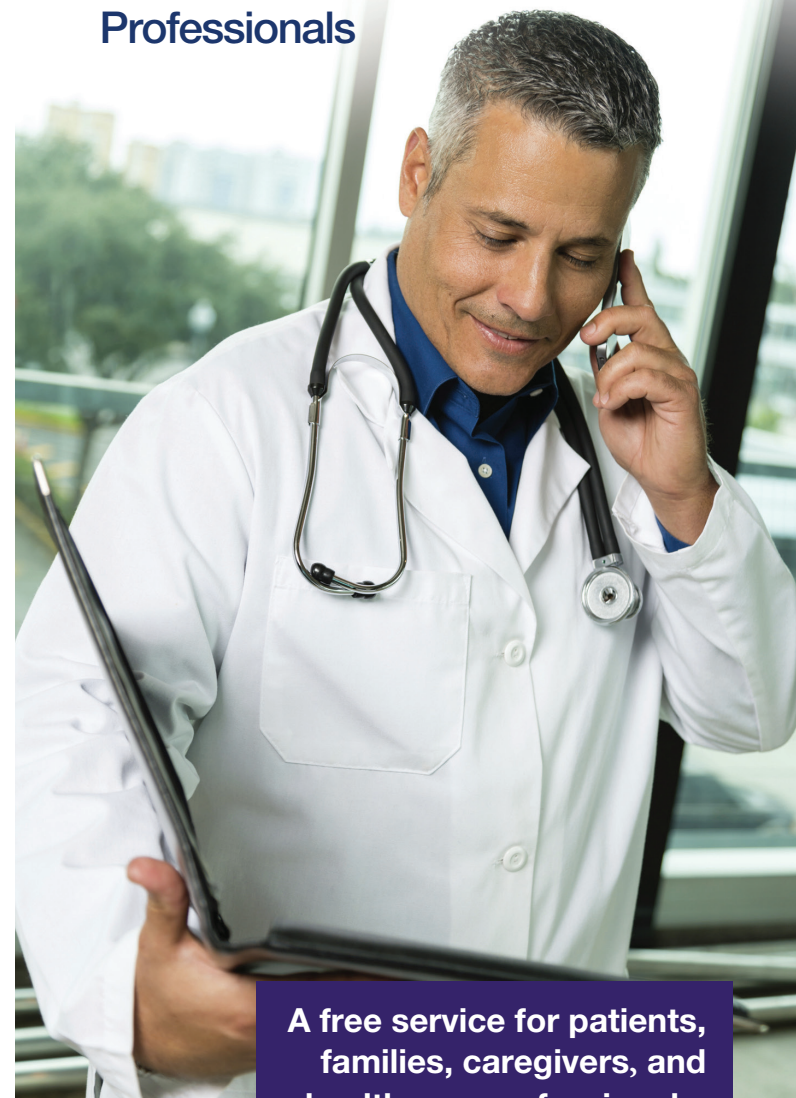
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# Alpha-1 Foundation Genetic Counseling Program

## A Guide for Healthcare Professionals



**A free service for patients,  
families, caregivers, and  
healthcare professionals.**



## About the Alpha-1 Genetic Counseling Program

The Alpha-1 Genetic Counseling Program provides free guidance for physicians and other healthcare professionals related to testing and diagnosis of Alpha-1 Antitrypsin Deficiency (Alpha-1). This service is provided via a toll-free number, 1-855-476-1227.

The program provides counseling, information, and referrals to resources for patients, family members, and caregivers.



**How Can I Contact the Genetic Counseling Program?**  
Call toll-free 1-855-476-1227

## What is Alpha-1 Antitrypsin Deficiency (Alpha-1)?

Alpha-1 is a genetic condition and one of the most common metabolic disorders. It follows co-dominant inheritance and can cause serious liver disease in neonates, young children and adults, or serious lung disease in adults. In some genetic variants, abnormal Alpha-1 Antitrypsin (AAT) protein builds up in the liver and cannot be released into the blood. Low AAT in the blood is unable to protect the lungs from injury, particularly from dust and cigarette smoke. In the absence of AAT, dust and cigarette smoke increase a chemical called neutrophil elastase that destroys the elastin supporting matrix of the lung, leading to Chronic Obstructive Pulmonary Disease (COPD).

## Testing for Alpha-1

The Clinical Practice Guidelines published in the Journal of the COPD Foundation, based on the latest evidence and six years of work, offer the following recommendations for Alpha-1 testing:

- Anyone who has COPD (emphysema and/or chronic bronchitis), regardless of age or ethnicity
- People who have unexplained chronic liver disease
- People who have necrotizing panniculitis, granulomatosis with polyangiitis, or unexplained bronchiectasis
- Parents, siblings, and children, as well as extended family members, of people who have been identified with an abnormal gene for Alpha-1, should be provided genetic counseling and offered testing for Alpha-1
- For family testing, alpha-1-protein-level testing alone is not recommended

because it does not fully characterize the risk of disease from Alpha-1

- For family testing or diagnostic testing of people who have symptoms, genotyping is recommended for at least the S and Z alleles. Advanced or confirmatory testing should include Pi-typing, alpha-1-protein-level testing, and/or expanded genotyping

To read the complete Clinical Guidelines, visit [a1f.org/clinical-guidelines](http://a1f.org/clinical-guidelines).

## Services for Healthcare Professionals:

- Information on inheritance and family risk
- Testing options
- Information on the pathogenesis of Alpha-1 in liver and lungs
- Treatment options
- Referrals to Alpha-1 Clinical Resource Centers
- Referrals to social services for your patients
- How to talk to your patient and family members about Alpha-1

## Confidentiality

The Alpha-1 Genetic Counseling Program provides information about genetic testing for Alpha-1, and the implications of an Alpha-1 diagnosis for your patients and their families. The privacy of all callers is protected to the extent permitted by law. Information you provide will not be shared with your patient, or with any organizations unless authorized by you.

The Genetic Counseling Program is operated by the Alpha-1 Foundation and complies with HIPAA regulations to protect the privacy and confidentiality of callers.