

Genetic Counselors

Genetic Counselors (GCs) are health professionals with a master's degree and experience in medical genetics and counseling. They augment 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.

GCs provide an additional resource for the healthcare professional. The expertise at the Alpha-1 Genetic Counseling Center 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. Genetic counselors can also give you specific guidance on the risks and benefits of genetic testing.

For more information about the Alpha-1 Foundation, visit our website at www.alpha1.org or call 1-877-228-7321.

The Alpha-1 Foundation Genetic Counseling Program is located at



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 has invested more than \$54 million to support Alpha-1 Antitrypsin Deficiency research at 100 institutions in North America, Europe, the Middle East and Australia.

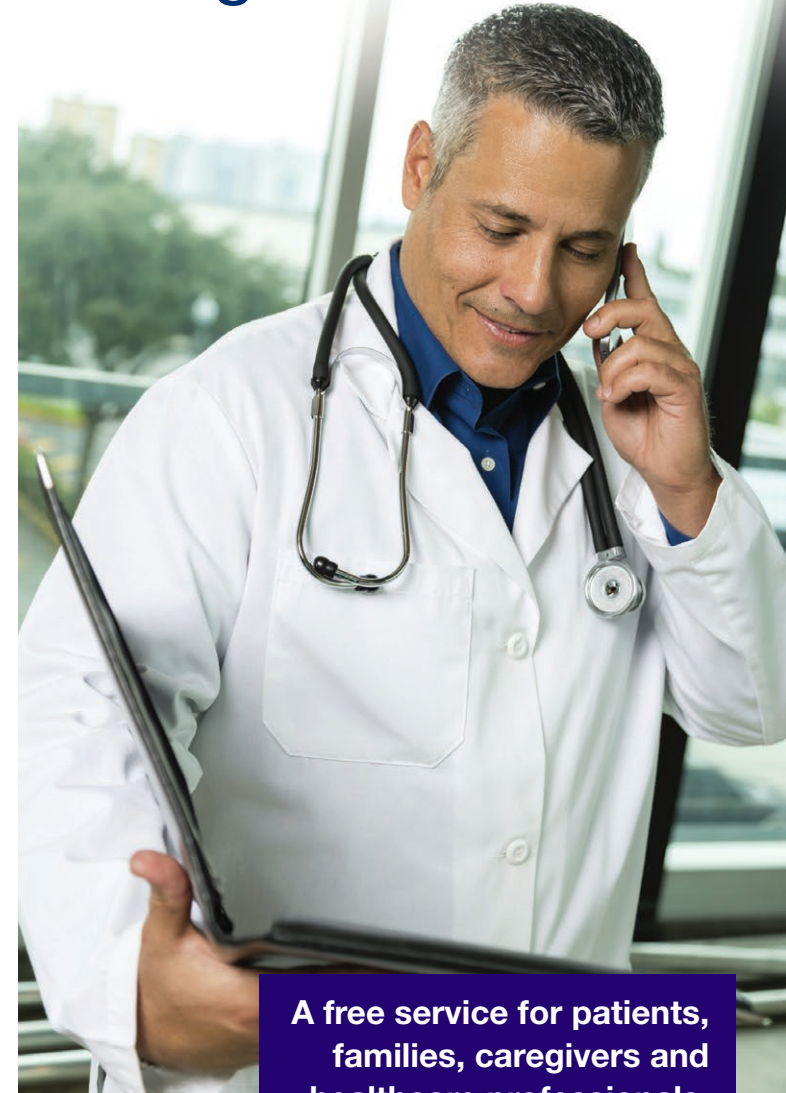
The Foundation also offers a wide array of programs for people diagnosed with Alpha-1, their families, caregivers and healthcare providers.

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

Unrestricted educational grants provided by:

Grifols

Alpha-1 Foundation Genetic Counseling Program



**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, 1-800-785-3177.

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 1-800-785-3177.

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 some 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 causing emphysema leading to chronic obstructive pulmonary disease (COPD).

Symptoms of Alpha-1

You may consider testing your patients for Alpha-1 if the following lung-related symptoms occur, or are a chronic condition:

- COPD
- Shortness of breath
- Wheezing
- Chronic bronchitis
- Bronchiectasis
- Recurring respiratory infections
- Non-responsive asthma
- Year-round allergies

Testing may also be recommended if these common symptoms associated with liver disease in Alpha-1 are present:

- Cirrhosis
- Unexplained liver problems
- Elevated liver enzymes

- Jaundice
- Ascites
- Vomiting blood
- Childhood failure to thrive

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 Antitrypsin Deficiency, 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 under the auspices of the Medical University of South Carolina, and complies with HIPAA regulations to protect the privacy and confidentiality of callers.