For More Information

For more information on the Alpha-1 Registry, please contact the ACT Trial Coordinator toll free at 1-877-886-2383 or email to "alphaone@musc.edu"

MUSC
MEDICAL UNIVERSITY OF SOUTH CAROLINA

Alpha-1 Research Registry
C/o Medical University of South Carolina
Division of Pulmonary and Critical Care Medicine
96 Jonathan Lucas St., Suite 812-CSB
PO Box 250623
Charleston, SC 29425
Toll Free Phone: 1-877-886-2383
Regular Phone: (843) 792-0260
Fax: (843) 792-0297
Email: alphaone@musc.edu

About the Alpha-1 Foundation

The Alpha-1 Foundation is a not-for-profit organization founded in 1995 by Alphas to promote research toward a cure for Alpha-1. Its mission is to provide the leadership and resources that will result in increased research, improved health, worldwide detection and a cure for Alpha-1. The majority of the Board of Directors is either diagnosed with Alpha-1 or is a family member of an individual diagnosed with Alpha-1. The Foundation has realized continuous growth since its inception and has developed a solid infrastructure to promote research and the development of new therapies for improving the quality of life for those diagnosed with Alpha-1.

ALPHA-1 FOUNDATION

Alpha-1 Foundation
2937 SW 27th Avenue, Suite 302
Miami, Florida 33133
Phone: 1-877-2-CURE-A1 (228-7321)
Fax: (305) 567-1317
Web Site: www.alphaone.org

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Family Linkage in the Alpha-1 Research Registry

Join the Alpha-1 Research Registry with your family.

Family Linkage in the Alpha-1 Research Registry

Now you can join the Alpha-1 Research Registry as a family! The purpose is to study genes that influence the development of disease in families with a history of Alpha-1 Antitrypsin Deficiency (AAT Deficiency or Alpha-1). Family members are invited to enroll if they carry the gene for AAT Deficiency or have the genetic disorder. Since genetic information privacy is an important issue, the Research Registry can now link family member records within the Registry database while protecting member confidentiality.

What is the Alpha-1 Research Registry?

The Alpha-1 Research Registry is a confidential database comprised of individuals diagnosed with Alpha-1 Antitrypsin Deficiency (Alpha-1) and individuals identified as Alpha-1 Carriers. The Registry was established by the Alpha-1 Foundation to facilitate research initiatives and promote the development of improved treatments and a cure for Alpha-1.

The Registry is located at the Medical University of South Carolina (MUSC) in Charleston. Individuals enrolled in the Registry have the ongoing opportunity to participate directly in clinical trials of new therapeutic approaches in addition to other research studies.

How can genetic studies benefit the Alpha-1 Community and lead to the development of new treatments and a cure for Alpha-1?

In the past scientists have been very successful at isolating genes commonly found in people with similar disease states. Once genes of interest are recognized, further studies can unlock the biological processes causing disease and ultimately lead to the development of new treatments. This is precisely the plan for genetic studies in Alpha-1.

It is known that the gene for the alpha-1 antitrypsin (AAT) protein plays a major role in the occurrence of AAT Deficiency. However, there are many important unanswered questions that genetic studies can shed light on. Such as why do some individuals with an abnormal Alpha-1 genotype never develop lung or liver disease and why do individuals with the same Alpha-1 genotype often suffer different severity of disease? Differences in environment such as cigarette smoking offer one explanation. However, it has been noticed that some people with similar Alpha-1 geneotypes have differences in lung or liver disease severity despite a similar smoking and environmental history. Another possibility for differences in the incidence and severity of Alpha-1 is the existence of other genes that affect the disease state. Such genes can become research targets to develop new therapies and treatments for AAT Deficiency.

Why are families so important for these studies?

Since families have a high percentage of identical genes, scientists can use small differences in a family's genetic code to narrow the range of genes that could be responsible for variability in the incidence and severity of Alpha-1. This alleviates the process of sorting through thousands of genes that make each individual unique. The goal of the Alpha-1 Foundation and Research Registry is to assist genetic investigators in the difficult task of finding study participants for these important family studies.

How are family member records linked confidentially?

If you are already a member of the Alpha-1 Research Registry, the Registry is asking you to speak with family members about joining the Registry as a "linked" family willing to participate in family studies. If you are aware of other family members who participate in the Registry please speak with them about linking your records. If you are not currently a member of the Research Registry please call the toll free number below and ask for Registry and Family Linkage applications.

The method to be used includes the following steps:

- **Fill out the Family Linkage Application.**
  Call or email the Registry for an application if needed. The Registry requires that you contact each listed family member and obtain his/her permission to be contacted by mail. *Note: The person who fills out the form for a family is known as the family index.*
- **The Registry will contact referenced family members requesting their participation and permission to be linked to other family members.**
  The index will not be informed of results of Registry contact with his/her family member(s). Even when families are linked in the Registry no family member will be able to request information concerning another family member's record or enrollment status.
- **Each family member will be presented with research studies separately and given the option to accept or decline the opportunity.**
  Some research will be as simple as filling out questionnaires or donating a blood sample and some research will be more involved. Not all studies are appropriate for all families for many reasons. Whenever possible the Registry will not reveal other family members' participation decisions. There will be some cases where this will not be possible. Please note, any family members can add or remove themselves from family linkage and/or the Registry at any time.

Joining the Registry as part of a family not only makes you eligible for family studies, but also for other studies the Registry supports. Notice of research opportunities and a biannual newsletter are sent through the mail. Please refer to the "Alpha-1 Research Registry, Be part of a cure" brochure for more information on the Registry.

Please carefully consider this opportunity. The goal of this project is to facilitate cutting edge genetic research that could answer complicated questions about the disorder and may eventually benefit people with Alpha-1. The Registry has tried to devise a method of linkage that is sensitive to family dynamics and alleviates family pressure, however there is no perfect solution. Every family that participates is helping to increase our understanding of AAT Deficiency. Please stress to family members the registry maintains strict procedures to maintain member confidentiality and no member is ever obligated to participate in any study. As always your support of Research Registry programs is greatly needed and appreciated.

Questions concerning family linkage will be answered at the Registry office from 8:30AM to 5PM Monday-Friday at 877-886-2383 or by email at alphaone@musc.edu.