

What Is The Alpha-1 Research Registry?

The Alpha-1 Research Registry, also located at MUSC, is a confidential database made up of people diagnosed with Alpha-1 and those identified as carriers of the disorder. The Registry was established by the Alpha-1 Foundation in order to facilitate research and promote the development of improved treatments and a cure for Alpha-1.

People in the Registry's database have the ongoing opportunity to participate directly in clinical trials of new therapeutic approaches in addition to other research studies. For more information about the Registry, please visit our website at www.alphaoneregistry.org or call 1-877-886-2383.

Please note: People who get tested through the ACT Study are not required to take part in the Alpha-1 Research Registry, but will be invited to participate if diagnosed with Alpha-1 or determined to be a carrier. They are not automatically added to the Registry database after receipt of their test results.



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.

www.alpha1.org
1 (877) 2 CURE A1 | 1 (877) 228.7321
3300 Ponce de Leon Blvd.
Coral Gables, FL 33134



Alpha-1 Coded Testing (ACT) Study

Confidential Testing for Alpha-1 Antitrypsin Deficiency



ACT study answers concerns about privacy of test results



How Can I or a Family Member Be Tested?

A study conducted through the Alpha-1 Research Registry at the Medical University of South Carolina (MUSC) under the direction of Charlie Strange, MD, provides a way for those at risk, including family members of already-diagnosed Alphas, to learn their Alpha-1 genotype. The genotype is the genetic code within all cells that determines physical characteristics governed by a particular gene.

The easiest way to get tested through the Alpha-1 Coded Testing (ACT) Study is to access the consent form online. Go to alphaoneregistry.org. From here you can click on “Get Tested” and

- Sign the electronic consent form that explains the research protocol
- Fill out the research questionnaire

If you click on “Learn More,” you can learn about the details of testing and our genetic counseling services. Or if no Internet access is available, paper forms of the consent and questionnaire can be mailed to you with a return envelope.

When the electronic forms (or paperwork) are completed and returned to MUSC, a coded,



finger stick blood test kit with directions for self-administration will be mailed. When the blood sample is returned to MUSC, it is then mailed for testing to GeneAidix Alpha-1 Antitrypsin Genetics Laboratory.

Results are sent to MUSC, linked with the participant and mailed to their home. In the event that a rare gene is detected during testing, they will be contacted by letter and invited to join a study by the Alpha-1 Foundation's DNA & Tissue Bank at the University of Florida.

Why “ACT” Now?

Many people at risk for Alpha-1 Antitrypsin Deficiency (Alpha-1) delay being tested for this genetic condition due to concerns about the privacy of test results and the possible effects on their future insurability. The Alpha-1 Foundation supports testing for people at risk because learning the results of these tests may begin a course of lifestyle changes and other modifications that could affect the onset and progression of the disease.

The Alpha-1 Foundation underwrites this confidential testing program based at MUSC. This free test is available through a research study, the Alpha-1 Coded Testing (ACT) Study, which investigates people's thoughts and feelings about the risks and benefits associated with learning genetic information.

Confidentiality

This program is being offered as a research study to gain knowledge about issues surrounding genetic testing. Participants' privacy will be protected to the extent permitted by law. All records are kept under lock and key and/or in a secure database. Finally, after receipt of results, a participant's name and other identifying information (such as an address) can be deleted from ACT Study records by request.

