



What Is The Alpha-1 Research Registry?

The Alpha-1 Research Registry, also located at the Medical University of South Carolina, is a confidential database made up of individuals 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 initiatives and promote the development of improved treatments and a cure for Alpha-1.

Individuals 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 contact the ACT Study coordinator. Please note, individuals who choose to participate in the ACT Study are not required to participate in the Alpha-1 Research Registry, but will be invited to participate if diagnosed with Alpha-1 or determined to be a carrier.



Research for a Cure

About the Alpha-1 Foundation

Alpha-1 Foundation is a not-for-profit organization dedicated to providing the leadership and resources that will result in increased research, improved health, worldwide detection, and a cure for Alpha-1 Antitrypsin Deficiency (Alpha-1). The leading experts in the field of Alpha-1 research are working with the Foundation through their participation on the Board of Directors, as members of the Medical And Scientific Advisory Committee (MASAC), directing Clinical Resource Centers, as members of Working Groups, or as participants at Foundation-sponsored scientific conferences and workshops. These experts, together with respected members of the Alpha-1 medical, professional, scientific, and patient communities are teaming up with the Alpha-1 Foundation to identify the most critical areas of research and support the development of new therapies. Importantly, the Alpha-1 Foundation has also formed collaborative relationships with government and industry to promote needed research and create awareness of this genetic disorder.

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Alpha-1 Coded Testing (ACT) Study

Confidential Testing for Alpha-1 Antitrypsin Deficiency



How Can I or a Family Member Be Tested?

A study conducted through the Alpha-1 Research Registry at MUSC under the direction of Charlie Strange, M.D., 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 which determines physical characteristics governed by a particular gene. Anyone who requests a test will receive a mailing that includes:

- Finger-stick blood test kit with directions for self-administration
- Consent form that explains the research protocol
- Research questionnaire



When the paperwork and blood test are completed and returned to MUSC, the blood test is coded with a unique research number. It is then mailed for testing to the University of Florida 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. For questions or to request a free test kit, please contact the ACT Study Coordinator toll free at 1 (877) 886-2383 or via email at alphaone@musc.edu.

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 a confidential testing program using the Medical University of South Carolina (MUSC). This 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.

