

**Medical University of South Carolina  
Electronic Consent to Be A Research Subject  
And HIPAA Authorization**

Title: Alpha-1 Coded Testing Study

Principal Investigator: Charlie Strange, MD

If participants include those under 18 years of age: 1) The subject's parent or legal guardian will be present when the informed consent form is provided. 2) The subject will be able to participate only if the parent or legal guardian provides permission and the adolescent (age 12-17) provides his/her assent. 3) In statements below, the word "you" refers to your child or adolescent who is being asked to participate in the study.

**A. PURPOSE AND BACKGROUND**

You have been identified because of your interest in obtaining a blood test for a protein called alpha-1 antitrypsin (Alpha-1). This document explains a way to receive that blood test through a research study designed to evaluate your thoughts and feelings about taking a blood test for a genetic disorder.

**B. PROCEDURES**

This study contains the following parts:

1. **You should read, sign, and date this consent form if you agree to participate.**  
Consent for minors less than 18 years of age should be from a legal guardian and also have the child's assent by signature if he or she is of age to understand why testing is being done. Once your consent and questionnaire (see #2 below) have been returned to us electronically, you will receive your Alpha-1 test kit in the mail.
2. Read and fill in the questionnaire. To receive your Alpha-1 test kit, you must submit a completed research questionnaire.
3. Obtain your blood sample by following the instructions in the blood drawing kit. You will receive a blood test kit that contains instructions for obtaining drops of blood, which will be collected on a piece of filter. The blood will be mailed to the Medical University of South Carolina (MUSC) where the sample will be coded with a research number and transferred to the University of Florida Health Science Center in Gainesville to test for Alpha-1. Results will be returned to MUSC. MUSC will link the coded research number with your address and mail you results of the test.
4. Mail the blood sample in the provided postage paid mailer.
5. By submitting this test, you also agree to receive future questionnaires that could arrive electronically or by mail at any time in the future. You also agree that ACT study staff can contact you for an updated email address, should your current email address become invalid.
6. If your finger stick test results suggest the possibility of a rare allele, a follow-up blood tube test by needle stick will be recommended since this is an important diagnosis. In

this event, you may request venipuncture tubes from the University of Florida DNA and Tissue Bank with a return address to Florida with results mailed to the participant and faxed to the ACT study.

#### C. DURATION

Your participation in the Alpha Coded Testing Study will be for life. Alpha-1 test results should arrive within 6 weeks of mailing.

#### D. RISK AND DISCOMFORTS

Blood obtained by finger stick involves puncture of the skin with a needle that can cause pain. Very rarely, infections can occur at the site of puncture. Blood obtained by needle stick involves puncture of a vein with a needle. Pain and bruising may occur at the needle site; rarely, infections can occur at the site of puncture.

The risks associated with obtaining a test for Alpha-1 include risks of establishing a new diagnosis. New diagnoses are often discovered by insurance companies and can cause insurance premiums to rise for you or your employer. New diagnoses that result from genetic tests can cause stress on relationships with family and friends. The questionnaires associated with this study will probe your feelings about testing and reasoning for testing.

Test results from the ACT Trial are returned to the participant through first class US Postal Service Mail only. MUSC cannot guarantee protection of your test results once they leave MUSC in the mail. It is a federal offense to open someone else's mail.

Although privacy protection procedures are in place, there is a chance of loss of confidentiality surrounding the test results.

Research to identify genes that cause or contribute to a disease or trait is an increasingly important way to try to understand the role of genes in human disease. You have been given this consent form because the Medical University of South Carolina investigators want to include your tissue, cell or blood sample in a research project, or because they want to save such biological samples for future research. There are several things you should know before allowing your tissues, cells or blood to be studied or to be stored.

1. Your tissue, cell or blood sample will be stored under an identifier which could be linked to you. Sometimes these samples are shared for research purposes with other investigators at other research sites. If this is done, the other investigators would not know your name.
2. In addition to your name, other information about you might be connected to your blood or tissue sample. For instance, information about race, ethnicity, sex, your medical history, and so forth might be available to investigators studying your tissue or blood. Such information is important for scientific reasons and sometimes for public health. It is possible that genetic information might come to be associated with your racial or ethnic group.
3. Genetic information about you will often apply (in one degree or another) to family members. It is not generally the University's policy to provide genetic information about you to your family members. However, certain studies, called "pedigree studies", share such information

among family members. For this and related research you will be asked if you are willing to share your genetic information with your family members.

4. You have the right to refuse to allow your tissue or blood to be studied or saved for future research studies. You may withdraw from this study at any time and remove any samples that contain identifiers from research use after the date of your withdrawal. This means that while the University of Florida might retain the identified samples-the law often requires this-they would not be used for research.

5. South Carolina law mandates that your genetic information obtained from any tests or from this research be kept confidential. Our state law prohibits any insurer using this information in a discriminatory manner against you or any member of your family in issuing or renewing insurance coverage for you or your family. Our state law further prohibits our sharing your genetic information with anyone except in a few narrow circumstances, one of these being a research project of this type, approved by the Institutional Review Board and then we must take all steps to protect your identity. You will still be responsible for paying for health care, however. The Medical University of South Carolina will not be responsible for such costs, even if care is needed for a condition revealed during research or clinical testing.

6. Genetic research raises difficult questions about informing you and other subjects of any results, or of future results. Some people feel anxious about the possibility of having a defective gene that would place them or their children at risk. Some people want to know what is found out about them; others do not. The risks of knowing include anxiety and other psychological distress. The risks of not knowing what is found include not being aware if there is treatment for the problem being studied. But these risks can change depending on whether there is a treatment or cure for a particular disease, and on how clear the results are. If there is a medical reason to seek specific information from you, your doctor will tell you this. A process called "genetic counseling" is often appropriate in such cases; you should ask your doctor or nurse about this if you have any questions.

Investigators in this study may try to recontact you in the future to find out about your health. If you are recontacted and want to know what the investigators have learned about your samples, you should understand that the following are the kinds of things the investigators or your health team might tell you:

- a) Information is too sketchy to give you particular details, but you will receive a newsletter informing you about the results of the project.
- b) You carry a gene for a particular disease that can be treated.
- c) You can carry a gene for a particular disease for which there is no current treatment. This news might cause severe anxiety or other psychological distress, depending on the severity of the disease.
- d) You carry a gene for a disease and might consider informing relatives that they, too, might carry the gene. It can be very difficult to decide whether to share such information with relatives. Genetic counselors can help sort out the various options in such a case.

Also, for any future research, we may contact you with a new consent form giving you additional information.

7. If you are concerned about a potential genetic disorder, you and your doctor might choose to test specifically for it. This would require additional blood or tissue samples and would not be part of this research project. You should discuss this option with your doctor or genetic counselor.

8. The presence of a genetic marker does not necessarily mean that an individual will develop a disease. Informing people of all such markers independently of medical need can cause unnecessary anxiety. On the other hand, the absence of a marker does not mean that someone will not get the disease. Genetic diseases appear as a result of a complex mixture of hereditary, environmental, behavioral and other factors.

These are the best-known risks and challenges of genetic research. There might be other risks we do not know about yet.

It is important that you talk to your doctor, nurse or genetic counselor if you have questions or concerns about the research study.

#### E. POSSIBLE BENEFITS

You will obtain an Alpha-1 blood test result that will determine if you have deficiency of Alpha-1. Severe Alpha-1 deficiency is sometimes associated with emphysema (lung damage) or liver damage that can be monitored and treated if you have this condition.

The test results will be returned only to you. Since you will be the only person that knows the result of your test, you can decide if you wish others to know this information.

The results of the questionnaires will be used to improve the care and support of Alpha-1 patients. You may obtain further information about Alpha-1 through this trial.

#### F. COSTS

There are no costs associated with this study; the test kit and results are free.

#### G. COMPENSATION

You will receive no compensation for participating in this trial.

#### H. ALTERNATIVES

You may have your physician send your blood to a different laboratory for Alpha-1 testing or elect to not be tested for Alpha-1.

#### I. CONFIDENTIALITY

To further help us protect your privacy, the investigators have obtained a Confidentiality Certificate from the Department of Health and Human Services.

With this Certificate, the investigators cannot be forced (for example by court subpoena) to disclose information that may identify you in any federal, state, or local civil, criminal, administrative, legislative, or other proceedings. Disclosure will be necessary, however, upon request of DHHS for audit or program evaluation purposes.

You should understand that a Confidentiality Certificate does not prevent you or a member of your family from voluntarily releasing information about yourself or your involvement in this research. Note, however, that if an insurer or employer learns about your participation and obtains your consent to receive research information, then the investigator may not use the Certificate of Confidentiality to withhold this information. This means that you and your family must also actively protect your own privacy.

Finally, you should understand that the investigator is not prevented from taking steps, including reporting to authorities, to prevent serious harm to yourself or others.

## **J. HEALTH INSURANCE PORTABILITY AND ACCOUNTABILITY ACT (HIPAA):**

Authorization to use or disclose Protected Health Information (PHI) for research Purposes.

HIPAA is a federal law that requires the protection of information that can identify you. Protected Health Information includes information that pertains to your past, present or future physical and mental health conditions, or the provision of health care. You are being asked to sign this Authorization because you are in the research study listed above.

The researchers agree to protect your protected health information by using and disclosing it only as permitted by you in the Authorization and as directed by state and federal law.

### **1. What is the purpose of the use and/or disclosure of your protected health information?**

You have submitted Protected Health Information to this study to assist in analyzing your blood sample for Alpha-1. No Protected Health Information will be voluntarily disclosed to individuals/entities outside the Medical University of South Carolina's (MUSC) Institutional Review Board for Human Research (IRB), which monitors human research at the university.

### **2. What protected health information will be used or disclosed?**

We will use the information submitted in the questionnaires to report on feelings about genetic testing, but you will not be identified. The blood card and attached questions are submitted to the lab at the University of Florida without information that can identify you. The MUSC IRB may request to audit this research study. We mail your test result to you. No PHI will be disclosed to anyone other than the parties above.

### **3. Who will disclose your protected health information?**

The Alpha Coded Testing Study Director and staff at the Medical University of South Carolina may disclose your medical/health information to the MUSC IRB if requested to monitor the study.

### **4. Who will receive your protected health information?**

You will receive your Protected Health Information in a letter containing test results that will be mailed to you. Your Protected Health Information may be shared with the MUSC IRB for purposes directly related to the conduct of the research. Once this information leaves MUSC, we cannot guarantee that it will be protected by this authorization.

### **5. Do you have to sign this authorization?**

You do not have to sign this authorization. If you choose not to sign the authorization, it will not affect your treatment, payment or enrollment in any health plan or affect your eligibility for benefits. You will not be allowed to participate in the research study.

**6. If you sign the Authorization, can you change your mind?**

You have the right to withdraw your authorization to allow MUSC to use or share your protected health information collected for this research study. Protected health information that has already been used or disclosed cannot be withdrawn. Your protected health information may still be used and disclosed if you have an adverse event. Once authorization is withdrawn and you are no longer participating in the study, no more protected health information will be collected. If you want to withdraw your permission, you must do so in writing to the investigator. The investigator's address is:

**Dr. Charlie Strange, MD.  
Medical University of South Carolina  
Division of Pulmonary, Critical Care,  
Allergy and Sleep Medicine  
96 Jonathan Lucas Street  
Suite 812-CSB, MSC 630  
Charleston, SC. 29425-6300**

If you withdraw your authorization, you will not be allowed to participate in the research study.

Results of this research will be used for the purposes described in this study. This information may be published, but you will not be identified. Information that is obtained concerning this research that can be identified with you will remain confidential to the extent possible within State and Federal law. The investigators associated with this study, and the MUSC Institutional Review Board for Human Research will have access to identifying information. All records in South Carolina are subject to subpoena by a court of law.

In the event of a study related injury, reasonable medical treatment not otherwise covered by third party payments or study sponsors will be available free through the Medical University (if approved by the South Carolina Budget and Control Board). Financial compensation is not available for medical treatment elsewhere, loss of work, or other expenses.

Your participation in this study is voluntary. You may refuse to take part in or stop taking part in this study at any time. You should call the investigator in charge of this study if you decide to do this. Your decision not to take part in the study will not affect your current or future medical care or any benefits to which you are entitled.

The investigators and/or the sponsor may stop your participation in this study at any time if they decide it is in your best interest. They may also do this if you do not follow the investigator's instructions.

**VOLUNTEER'S STATEMENT**

I have been given a chance to ask questions about this research study. These questions have been answered to my satisfaction. If I have any more questions about my participation in this study or study related injury, I may contact Charlie Strange, MD or his research associates at 1-877-886-2383. I may contact the Medical University of SC Hospital Medical Director (843/792-9537) concerning medical treatment.

If I have any questions, problems, concerns, desire further information or wish to offer input, I may contact the Medical University of SC Institutional Review Board for Human Research IRB Manager or the Office of Research Integrity Director at (843) 792-4148. This includes any questions about my rights as a research subject in this study.

Your typed name here indicates your agreement to participate in the ACT study.

What is today's date?

What is your email address?

What is your age?

(IF PARTICIPANT IS UNDER 18:)

If the participant is under 18 years old, your typed name here indicates your agreement for your child to participate in the ACT study.

What is today's date?