Summary Alpha-1 Antitrypsin Study

All healthy humans have many micro-organisms (“germs”) in their upper respiratory tract, gastrointestinal tract, and some other parts of the body such as the skin. These micro-organisms are important in a variety of normal biologic processes. However, we think that the numbers and types of these micro-organisms may change in the lungs in people who have Alpha-1 Antitrypsin Deficiency (AATD, Alpha-1) and that the changes may affect the course of the disease. The purpose of this research study, called Genetic Research in Alpha1-Antitrypsin Deficiency and Sarcoidosis, GRADS for short, is as follows:

• We want to identify the types of micro-organisms in the lungs and other tissues in people with Alpha-1 or the carrier state (individuals who carry one abnormal Z gene).
• We want to examine the blood of patients with Alpha-1 for factors that indicate swelling, or inflammation.
• We want to compare the types of micro-organisms present in the lung with the inflammatory patterns in the blood and with symptoms of Alpha-1 lung disease.

If you agree to participate in this study, you will see the researchers and coordinators two times over the course of 1 month. At those visits, the following will occur:

Visit 1(can be completed in 1 or 2 days)

• Explanation of all study procedures
• Sign consent form
• Complete history and physical examination
• Blood, Urine and stool samples
• Questionnaire
• Pulmonary function test
• Six minute walk test
• Pregnancy test
• CT scan (this can be done within the first 3 weeks)

Total number of minutes for completion of the first visit is 196 min ~3.5 hrs (whatever is not completed on the first visit can be completed in the second visit)

Visit 2

• Bronchoscopy and broncho alveolar lavage
• Tongue scraping
• Bronchial brushing (if agreed)

The bronchoscopy procedure will take about 45 minutes, but you will not be discharged until anesthesia has worn off and your breathing is back to normal.