

## 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.