

5000 COPD Patients Tested for Alpha1 Antitrypsin Deficiency

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DENVER —

Underdiagnosed, inherited form of the disease has different therapy

National Jewish Medical and Research Center pulmonologist [Robert Sandhaus, MD, PhD](#), will lead a study seeking to identify patients who have an inherited form of chronic obstructive pulmonary disease (COPD), known as [alpha-1 antitrypsin deficiency](#) (Alpha-1). Those patients have a better prognosis than most because they can receive therapy that can slow the progression of the disease. The study, sponsored by the [Alpha-1 Foundation](#) and the [American Association for Respiratory Care](#), is seeking more effective ways to identify Alpha-1 patients.

"If this study confirms what much smaller studies have suggested about Alpha-1 patients hidden in the COPD population, there could be as many as 400,000 individuals in the US who have lung disease due to Alpha-1, don't know it, and aren't getting optimal therapy," said Dr. Sandhaus, who is also Clinical Director of the Alpha-1 Foundation. "We are trying a new model, using respiratory therapists to identify Alpha-1 patients, which we hope will greatly increase diagnosis of alpha-1."

More than 12 million people in the United States have been diagnosed with COPD, an umbrella term that includes emphysema and chronic bronchitis. COPD is the fourth leading cause of death in the United States. Alpha-1, sometimes called "genetic COPD," is the most common known genetic risk factor for emphysema.

Alpha-1 can be detected with a simple finger stick test. The American Thoracic Society, American College of Chest Physicians, European Respiratory Society, and the American Association for Respiratory Care all recommend testing all symptomatic COPD patients for Alpha-1. However, physicians have been slow to adopt those recommendations. Respiratory therapists, who administer routine lung-function tests to COPD patients, have enthusiastically supported such testing, said Dr. Sandhaus.

So, Sandhaus and his colleagues will test and evaluate a program to have respiratory therapists ask all COPD patients referred to them for lung-function testing if they want to be tested for alpha-1.

"The expanding role of the RT in supporting the physician will accelerate early detection and access to appropriate care," said Alpha-1 Foundation President and CEO John W. Walsh.

"The positioning of respiratory therapists in this process is a natural relationship," Thomas Kallstrom, AARC Chief Operating Officer. "We're proud to be part of this monumental study."

In addition to determining the prevalence of undetected alpha-1 antitrypsin deficiency, the study will seek to identify non-genetic parameters of lung function that are likely to predict the presence of Alpha-1, and the age distribution of positive testing for Alpha-1.

The study will enroll 5,000 people at 15 academic sites around the United States. To be eligible, a patient must have been referred for pulmonary function testing at the site. When the pulmonary function testing is complete, if the patient meets COPD criteria (as defined by the Global Initiative for Chronic Obstructive Lung Disease) the patient will be offered enrollment in the study.

Those who decide to participate will be given a finger-stick test for Alpha-1 and will complete a brief questionnaire. The study requires only about 30 minutes of the subject's time.

Participants will receive the results of their testing. If they are positive for Alpha-1, they will receive educational information about the condition and will be encouraged to call a toll-free number to speak with the Alpha-1 genetic

counselor at the Medical University of South Carolina.

To evaluate the relationship between age and Alpha-1 diagnosis, the study will include equal numbers (1,667 per age group) of three age groups: 18-50; 51-65; and greater than 65 years old.

The epidemiology and AAT genotyping for the study will be done at the University of Florida Alpha-1 Genetics Laboratory run by Mark L. Brantly, MD. Data coordination and statistical analysis will be done by James Murphy, PhD, at National Jewish.

Jorge Zamudio, MD, is the Alpha-1 Foundation coordinator.

The study is supported in part by an unrestricted charitable contribution from Talecris Biotherapeutics.

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