

The National Survey of Patients with Alpha-1 Antitrypsin Deficiency

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To better define issues affecting the Alpha-1 community, the Alpha-1 Association, the Alpha-1 Foundation, and AlphaNet commissioned a joint national survey in the spring of 2003 to focus on patient perspectives concerning healthcare and Alpha-1 community issues. The survey was conducted through Schulman, Ronca, and Bucuvalas, Inc. (SRBI), an international research organization in Washington, DC. SRBI signed a confidentiality agreement with each of the three sponsoring organizations so that they could construct a single sampling population from the mailing list of all three organizations omitting duplicates. From these lists there were a total of 5,222 unique potential participants representing the best estimate of the number of diagnosed persons with Alpha-1 Antitrypsin Deficiency in the US. Questionnaires were sent to the participants beginning in April 2003. By June 11, 2003 1,953 (37%) questionnaires had been returned to SRBI.

Results showed severe deficiency of AATD (PiSZ, PiZZ, Pi Znull) was present in 92.7% of the survey participants. Another 2.0% were caregivers answering on behalf of a deficient individual and 4.8% were PiMZ carriers. Carriers were excluded from the remainder of the analysis unless specifically reported. The mean age of participants was 53.1 years. We compared the percent of Alpha-1 respondents in each age category to the general population based on the 2000 US census. As expected, results show the majority of Alpha-1 patients are middle-aged as symptoms most often present after the third decade. Sixty two percent of known Alpha-1 patients are age 45-64, while only 22% of the general population falls into that age range. Ninety percent of the survey population falls within the age range 35-75, while 45% of the US population falls in that age range. Few children and young adults are known in the Alpha-1 community since the majority of symptoms present later in life.

The mean age at first diagnosis of AATD was 43.9 years. The majority of participants (58%) were diagnosed between ages 40-59, 9% were diagnosed between ages 60-79 and less than 1% were diagnosed after age 80. More than a third (40%) of respondents report being diagnosed within a year after seeking treatment for

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symptoms. Overall the mean time between seeking treatment for symptoms and diagnosis was 5.6 years. It seems that physician knowledge of Alpha-1 in the US may be improving as time to diagnosis decreased as decade of birth increased. This suggests the awareness and Alpha-1 education efforts of national patient organizations have been effective. For those born before 1950 the mean time between seeking treatment for symptoms and diagnosis was 7.0 years. The mean time to diagnosis decreased to 4.5 years for those born in the 1950s and 4.0 years for those born in the 1960s.

Participants were able to self-report any combination of diagnoses associated with Alpha-1. The prevalence of emphysema, asthma, chronic bronchitis, and liver disease in the survey population was 53%, 45%, 38% and 8% respectively. Sixteen percent of respondents do not report any pulmonary or hepatic symptoms. Among Alpha-1 carriers, 46% did not report any symptoms, 33% report asthma, 29% report chronic bronchitis, 15% report emphysema, 14% report COPD and 8% report liver disease. Although Alpha-1 is classically characterized as a cause of genetic emphysema, this survey highlights the high prevalence of wheezing, cough, and bronchitis in this population. The clinical message is that an Alpha-1 test should be ordered for every patient with COPD as suggested in the recent ATS/ERS statement.

Sixteen percent of severely deficient Alphas report having had a transplant or being on a transplant list. Those participants report age at first diagnosis to be significantly younger (group mean 39.7 years versus 45.2 years) than Alphas who do not report a transplant or being on a transplant list. This suggests that persons diagnosed in the third decade of life, presumably due to symptoms, may be more likely to need a transplant in the future than persons diagnosed in the fourth decade of life or later.

Nearly three quarters (74%) of the severely deficient Alphas report that they are currently infusing augmentation therapy. Participants infuse less frequently than scheduled often (3%), sometimes (25%), rarely (50%) and never (22%) in the respective percentages. Overall, the majority (62%) of non-carriers say they would be willing to try another FDA approved IV therapy at the same cost. When compared to participants not on augmentation therapy, those reporting augmentation therapy use are older, diagnosed at an older age, report longer time to diagnosis after onset of first symptom, are more likely to report symptoms of lung disease, more frequently use oxygen therapy, and report better quality of life.

Survey responses show that most participants were familiar with and satisfied with the three main national Alpha-1 patient organizations; the Alpha-1 Association, the Alpha-1 Foundation and AlphaNet. In particular the respondents look to these organizations to promote development of new therapies, research funding, physician education, screening and detection, and public awareness. Notably over 90% of respondents rate their AlphaNet Coordinator as excellent or good in providing service, information and support. Respondents felt that COPD education is an appropriate topic for Alpha-1 Education Days.

In conclusion, this survey tells us that less than half of the known Alphas in the United States are members of the Alpha-1 Research Registry as there 2,500 Registry members and there were 5,222 individuals eligible to receive the SRBI survey. Overall, participants are familiar with the national patient organizations and support their activities. This survey provides some evidence that early diagnosis provides benefits to the population and suggests that screening for this disorder in all patients with COPD should be routinely performed.

