



## ALPHA-1 ANTITRYPSIN DEFICIENCY: HEALTH AND PSYCHOSOCIAL CHALLENGES FOR PATIENTS/CAREGIVERS IN CANADA SURVEY SUMMARY REPORT 2007

In the summer of 2007, Alpha-1 Antitrypsin Deficiency Canada Inc. (Alpha-1 Canada), commissioned a survey to capture the ongoing challenges of patients with Alpha-1 Antitrypsin Deficiency (A1AD).

### Survey Highlights

- Patients have experienced significant delays in both diagnosis and treatment of their disease. Lung disease is the primary challenge of patients who are afflicted with A1AD, with liver and skin disease being less common. For patients with lung disease, the mean age of diagnosis was 45.5 years with a delay in diagnosis reported as 9.9 years. Although only a limited number of respondents (10%) reported symptoms of liver disease, symptoms developed an average of 4.5 years prior to A1AD diagnosis. Following diagnosis, 40% of the patients experienced a delay in the initiation of treatment (mean delay of 4.2 years).
- Misdiagnosis of A1AD over the last two decades has contributed to the premature loss of lung function and deterioration in overall health status of patients. A1AD lung patients (28%) reported suffering with asthma for an average of 14 years prior to obtaining a definitive diagnosis for their respiratory disease.
- A1AD patients rely heavily on health care services to manage their symptoms. In the course of the last year, patients reported an average of 3 visits to family physicians and 2.7 visits to specialists to treat A1AD symptoms. In addition, 22% of respondents had sought emergency room services and 10% were hospitalized in the last year.
- Financial barriers prevented A1AD patients from following treatment recommendations prescribed by physicians. Even though augmentation therapy was considered a treatment option for A1AD patients with lung disease, 36% of the patients who were prescribed the therapy did not receive it. Reasons for not receiving the prescribed treatment included i) lack of reimbursement by government or private insurance, ii) inability to pay the out of pocket costs and iii) a patient's decision not to initiate therapy.
- A1AD adversely affects patients in the prime of their life, impacting both employment and social interactions with friends and family. More than half (56%) of the respondents indicated that it adversely affected their employment, either causing them to change jobs or reduce their work hours. Other adverse social effects included decreased interactions with family, greater feelings of isolation, and significant limitations on recreational activities.
- Many A1AD patients face barriers, including discrimination, in obtaining health, disability and life insurance coverage.
- Provincial health care programs and services for A1AD patients are not funded consistently throughout Canada. There are major differences in funding of diagnostic tests, such as phenotyping, and treatment such as augmentation therapy.

This survey demonstrates that A1AD patients experience significant health, psychosocial and financial burdens. To improve the disease management of A1AD, areas of focus should include: (1) broader education in primary health settings to ensure early identification and proper diagnosis, and prompt initiation of treatment, (2) review of national/provincial funding of diagnostic tests and treatment options to ensure equitable access to all Canadians, (3) a challenge to insurance companies regarding genetic discrimination, and (4) further develop patient support programs to provide up to date education and help to cope with the psychosocial implications of the disease.