European Respiratory Society
Annual Congress 2012

Abstract Number: 4891

Publication Number: P2273

Abstract Group: 5.2. Monitoring Airway Disease

Keyword 1: COPD - diagnosis **Keyword 2:** Genetics **Keyword 3:** Chronic disease

Title: Trends in diagnosis and clinical presentation of alpha-1 antitrypsin deficiency within an Irish population

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Body: Alpha-1 Antitrypsin Deficiency (AATD) is an autosomal co-dominant genetic disorder associated with a substantially increased risk for the development of chronic obstructive pulmonary disease (COPD) and liver disease. AATD is a notoriously under-diagnosed and under-recognized condition. ATS/ERS guidelines recommend testing of all individuals with COPD and poorly controlled asthma. The objective of the study was to investigate the diagnostic experiences of ZZ AATD individuals in Ireland. A total of 50 ZZ AATD individuals completed a questionnaire at an Alpha-1 Clinic in relation to their diagnostic experiences and clinical presentation. The mean age of symptom on set was 36.7 years +/- 1.7 (range 4-60); mean age of diagnosis was 42.9 years +/- 1.6 (range 4-68). The interval between onset of symptom and diagnosis was 6.2 years. The mean number of physicians seen prior to a diagnosis was 2.6 +/- 0.3 (range 1-13). Symptomatically screened ZZ individuals mean age of symptom on set was 36.2 years+/- 2.0 (range 4-60): mean age of diagnosis was 44.3 years +/- 1.9 (range 4-68). The interval between onset of symptom and diagnosis was 8.1 years. The number of physicians seen prior to a diagnosis was 3.1 +/-0.3 (range 1-13). Family screened ZZ individuals mean age of symptom onset was 37.63 +/- 3.0 (range 21-51); mean age of diagnosis was 38.5 +/- 2.8 (range 21-53). The interval between onset of symptoms 0.9 years. The mean number of physicians seen prior to a diagnosis was 1.2 +/- 0.2 (range 1-3). Our results further underline the need for increased awareness and early detection of symptomatic AATD individuals in the Irish population, especially among the COPD popluation.