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Title: Characteristics of zz alpha-1 antitrypsin deficiency patients on the Irish national registry

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Body: Rationale: Alpha-1 antitrypsin (AAT) is produced by hepatocytes, and is the most important antiprotease in the lung. AAT deficiency (AATD) is a hereditary disorder resulting from mutations in the AAT gene. Individuals with this deficiency classically present with lung disease in adulthood. WHO guidelines advocate a targeted strategy in screening COPD, non-responsive asthma, cryptogenic liver disease patients and relatives of known AATD patients. Methods: The most common AAT phenotype associated with lung disease is ZZ. A chart review of AATD patients on the National Alpha-1 Registry was performed on ZZ patients (n=100). Our registry collects data on pulmonary function tests, GOLD guidelines, initial reason for screening, complications, and smoking history. Results: We found that ZZ individuals identified as a result of family screening have significantly increased FEV1 (85.3 +/- 6.5%, 40.8 +/-2.8 years) compared to ZZ patients identified by targeted symptomatic screening (54.38 +/-3.99%, 44.86 +/-1.8 years, p=0.0008). ZZ patients with a history of smoking had significantly decreased lung function (FEV1, 54.8 +/- 3.9%, 43.71 +/- 1.6 years) compared to never-smoking ZZ individuals (FEV1, 88.24 +/- 4.8%, 43.39 +/- 3.6 years, p<0.0001). Conclusions: Our results highlight the role of cigarette smoke in the pathogenesis of lung disease in AATD and the need for increased awareness and early detection of asymptomatic AATD. Identification of patients from a targeted detection programme should include aggressive family screening and allow the initiation of preventative measures before significant lung disease has occurred.