8.04 Evaluating the benefits of family screening in severe AATD

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Background: Alpha-1 antitrypsin deficiency (AATD) is a genetic disorder that can manifest as lung, liver and rarely skin disease. Smoking is a major risk factor in lung disease onset and severity. This study aimed to investigate family screening benefits in severe AATD and to demonstrate smoking impact on the lungs. **Methods:** Participants on the Irish National AATD Registry completed a questionnaire on demographic information, including smoking and occupational history. Pulmonary Function Tests (PFT) at diagnosis and Computed Tomography (CT) reports were collected to evaluate lung function impairment and structural damage, respectively. **Results:** Of the participants (n=261), 36.4% were detected by family screening. Individuals in the family screening cohort had less severe obstruction on spirometry and less likely to have radiographic emphysema. A significant difference in FEV1% predicted was found between ever-smokers and never-smokers (p=0.00001). The odds ratio (OR) of having either emphysema (p=0.00001) or combined emphysema-bronchiectasis (p=0.00001) was significantly raised in ever-smokers. Mean age at death(n=44) was 70 in never-smokers compared to 61 in ever-smokers (p=0.0221). **Conclusion:** Early identification of at-risk individuals with severe AATD through family screening would allow smoking cessation interventions, and lead to less lung disease in this highly susceptible population.
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