A 76 Year Old Female Diagnosed with Cystic Fibrosis

Abstract:
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The diagnosis of Cystic Fibrosis (CF) requires a high clinical suspicion in patients presenting at all ages. Early recognition permits referral to a specialist centre and may reduce the morbidity and mortality associated with CF. We report the case of the oldest patient in Ireland diagnosed with CF at 76 years of age and highlight the clinical features of her presentation.

Introduction
Cystic Fibrosis is the most common fatal inherited genetic condition in Ireland with a carrier frequency of 1:18 and incidence of 1:153, the highest worldwide. The pathogenic manifestations of CF are due to reduced CFTR-related chloride conductance, this leads to airway surface liquid depletion, reduced mucociliary clearance, and retention of secretions, thus promoting recurrent infection and chronic inflammation. The majority of patients with CF are diagnosed in childhood, but recently this is becoming rarer, with the recognition of milder phenotypes, more prevalent in those diagnosed with CF in adulthood (>40 years). CF patients diagnosed in adulthood had similar lung function decline, and died from similar causes, compared to those diagnosed in childhood. Importantly, this highlights the morbidity of CF lung disease in this group and that it does not run a benign course.

Case Report
A 76-year-old woman was referred for assessment of possible bronchiectasis. She reported a history of recurrent childhood pneumonia, asthma and had a remote smoking history. She had a daily cough productive of yellow sputum and frequent hospital admissions with lower respiratory tract infections. On examination, her body mass index was 28kg/m², and auscultation of her chest was clear. Pulmonary function testing demonstrated normal spirometry values with an obstructive ratio: FVC 2.29 (106%), FEV1 1.59L (93%), FEV1/FVC 69%. An initial chest radiograph was reported as normal, her chest x-ray showed a right middle lobe bronchiectasis (Figure 1) with otherwise normal lung parenchyma. Bronchoalveolar lavage cultured Stenotrophomonas maltophilia, Haemophilus influenza and Staphylococcus aureus. Repeat sputum cultures isolated a non-tuberculous mycobacterial (NTM) species, Mycobacterium chelonae. Sequential sweat chloride testing was positive with values of 62mmol/L and 63mmol/L (>60mmol/L; CF highly likely).

Discussion
The natural history of CF has evolved considerably in recent decades; improvements in airway clearance, nutrition and antibiotic therapy have led to significant increases in life expectancy. The diagnosis of CF in adulthood (adult CF) is a relatively uncommon event, with the median age of diagnosis of 11 years old. In Ireland, the oldest patients with CF are those diagnosed in childhood. Importantly, this highlights the morbidity of CF lung disease in this group and that it does not run a benign course.

Our patient had many of the characteristics identified by this study; female sex, pancreatic sufficiency, NTM-culture positive sputum and evidence of CFTR dysfunction. Her preserved lung function highlights the potential role of gene modifiers. At 76 years of age, she is the oldest patient ever to be diagnosed with CF in the Republic of Ireland and she remains stable at 78 years of age. The diagnosis of CF in this patient permits her care to be coordinated by our adult CF centre, including direct access to the multidisciplinary healthcare structure. She attends for three-monthly outpatient follow up, receives earlier and more aggressive antibiotic therapy for pulmonary exacerbations, and genetic counselling has been provided to her family. Our case report highlights that in those with appropriate clinical findings, investigations to exclude CF should be instigated at any age. The presence of typical CF or NTM pathogens in sputum would further alert the clinician to possible underlying CF. Adults with CF are a rapidly ageing cohort, and as illustrated by our case, some with milder phenotypes can expect to live a normal lifespan.

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