Cystic fibrosis in a 70 year old woman

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Abstract
A 68 year old woman with a lifelong history of chronic bronchitis was diagnosed as having cystic fibrosis. The diagnosis was based on a suggestive family history, steatorrhea, bronchiectasis with respiratory insufficiency, and very high sweat sodium content. The patient was found to be heterozygous for the delta F 508 gene defect.

Cystic fibrosis is regarded as a disease of childhood and more than 95% of patients have the diagnosis made in childhood. We present a woman with a long history of respiratory infections who was diagnosed as having cystic fibrosis at the age of 68 years. This case illustrates that cystic fibrosis should be considered in all age groups.

Case report
The patient, a Dutch woman, was born in 1920. From early childhood she had suffered from a cough, producing thick, purulent sputum every day and having episodes of haemoptysis. Her symptoms diminished in her 20s but she still occasionally had attacks of bronchitis. At the age of 36 she underwent a cholecystectomy. After the age of 50 her pulmonary symptoms and function worsened and she frequently required medical treatment. During the last two decades Staphylococcus aureus and Haemophilus influenzae were isolated repeatedly from her sputum. Pancreatic insufficiency was shown by a high fat loss in her stools, but she had no symptoms of malabsorption and needed no pancreatic enzyme substitution. Over the years her pulmonary function declined, finally resulting in the use of corticosteroid and supplementary oxygen treatment at home. In 1988, at the age of 68 years, she was admitted to our department because of an exacerbation of dyspnoea and a productive cough. Three of her sisters had died, aged from 6 weeks to 6 months, because of "sticky mucus." She is married, has uncomplicated pregnancies, and has two healthy sons. Her children and grandchildren have had no respiratory or gastrointestinal symptoms. Physical examination showed a 68 year old well nourished woman (height 163 cm, weight 65 kg). Her pulse rate and blood pressure were normal. The nasal passages were clear and there were no signs of chronic sinusitis, clubbing, or ankle oedema. Coarse crackles were heard over both lungs and especially in the upper parts. Heart sounds were normal and abdominal examination showed no abnormality.

Glucose and haemoglobin concentrations and results of liver and renal function tests were normal. The fat soluble vitamin concentrations were low. Arterial blood gas analysis showed that the pH was 7.36, oxygen tension (PaO₂) 8-4 kPa, carbon dioxide tension (PaCO₂) 7.8 kPa, and oxygen saturation (SaO₂) 91% while she was having 1.5/min oxygen. Chest radiograph (figure) showed peribronchial thickening, bronchiectasis, and reduction of the volume of the upper lobes. A sinus radiograph showed no evidence of sinusitis.

Pulmonary function tests showed a combined restrictive and obstructive defect (vital capacity 0.99 l (37% of predicted) FEV₁ 0.40 l (19% of predicted, with no reversibility after bronchodilator).

Haemophilus influenzae and late Pseudomonas aeruginosa were isolated from sputum. Faecal fat excretion was about 53 g over 72 hours with a fat intake of 240 g (normal loss is 2 g/day). The sweat sodium concentration (the patient had been taking 10 mg oral prednisone daily for many years) was 94 mmol/l for both the right and the left arm in samples of respectively 106 and 139 mg of sweat.

This combination of findings and a suggestive family history suggested the diagnosis of cystic fibrosis.

The patient proved to be a compound heterozygote with the genotype delta F508/unknown. Other probes tested were G542 x, d I 507, G 551 d, S 594 N and S 549 I. Other family members refused genetic analysis as they had no complaints.

She was treated with antibiotics, oxygen, and chest physiotherapy. Over the past three
Discussion
Cystic fibrosis is an autosomal recessive genetic disorder. Recently the cystic fibrosis locus has been located on chromosome 7 and the nucleotide base sequence of the gene has been determined. The most frequently reported mutation is delta F508.1 Cystic fibrosis includes chronic obstructive and suppurative lung disease, exocrine pancreatic deficiency and abnormally of the liver and of the reproductive tract.2,3 The diagnosis is based on the detection of increased concentrations of electrolytes in the sweat.4 There is often a family history of cystic fibrosis. Expression of the disease varies and there is a little relation between age at diagnosis and age at death. A few elderly patients with cystic fibrosis have been reported,5,6 but our patient is probably the oldest to be diagnosed and reported. This case shows that in patients with appropriate symptoms cystic fibrosis should be considered whatever their age. A mild clinical course may delay diagnosis and may be related to heterozygosity.

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