CASE STUDY

ADULT CYSTIC FIBROSIS

John Paul Clancy, MD

BACKGROUND
At the time of presentation, CB was a 38-year-old Caucasian male who was referred to the adult pulmonary tertiary care clinic with an increasing history of hemoptysis. Symptoms were noted for approximately 3 months, with occasional sputum production that had increased from blood streaking of sputum to expectoration of frank blood several times per week. A chest X ray performed by his local physician was concerning for possible bronchiectasis, prompting referral to a tertiary care center.

PAST MEDICAL HISTORY
The patient’s past medical history is significant for chronic sinusitis and occasional bronchitis, successfully treated with oral antibiotics.

FAMILY HISTORY
There is a family history of diabetes and hypertension, with no other identified lung, gastrointestinal, pancreatic, or hepatic disease.

PERSONAL AND SOCIAL HISTORY
CB has been married for over 10 years and has no children. He works as a private businessman. The patient had been a college football player (offensive lineman), and had required intravenous fluid administration on several occasions for hyponatremic dehydration during his college football experience.

REVIEW OF SYSTEMS
Review of systems is negative for other abnormalities, including weight loss, diarrhea or constipation, gastroesophageal reflux, fevers, rashes, or other systemic findings.

PHYSICAL EXAMINATION
CB’s physical examination was significant for mild obesity (weight 110 kg) with normal heart rate, blood pressure, and temperature. The physical findings of note included occasional pulmonary crackles in the upper lung fields without respiratory distress. No clubbing was identified. His head, ears, eyes, nose, throat, cardiac, abdominal, skin, musculoskeletal, neurologic, and cognitive examinations were otherwise unremarkable.

DIAGNOSTIC STUDIES
A chest X ray performed in clinic revealed bilateral bronchiectasis in the upper lung fields. Pulmonary function (spirometry) tests revealed a mild obstructive pattern, with a forced vital capacity of 96% predicted, forced expiratory volume in 1 second (FEV₁) of 70% predicted, and forced expiratory flow between 25% and 75% of forced vital capacity of 53% predicted. A sputum culture grew Pseudomonas aeruginosa with a mucoid phenotype, and sweat testing revealed elevated sweat chloride values of 82 µmol and 80 µmol on sequential analysis. Genotyping identified the heterozygous presence of the ΔF508 mutation in cystic fibrosis transmembrane conductance regulator (CFTR; 24 mutation screen). Metabolic profile, complete blood count, vitamin levels (A, D, and E), and clotting studies were normal, and no evidence of pancreatic insufficiency was identified.

DIAGNOSIS AND TREATMENT
Based on the findings of bronchiectasis, positive sweat chloride testing, and sputum cultures positive for P aeruginosa, the diagnosis of cystic fibrosis (CF) was made. The patient was hospitalized and treated with intravenous antibiotics (ceftazadime and tobramycin) for 10 days with regular chest physiotherapy, nebulized dornase alpha, and bronchodilators. Extensive education was completed by the CF care team. At the completion of his inpatient treatment, CB’s FEV₁ increased to 82% of predicted, with resolution of his ongoing hemoptysis. He was discharged home on daily chest physical therapy, daily dornase alpha therapy, and use...
of bronchodilators as needed, with regularly scheduled CF clinic visits at 3-month intervals.

**LONG-TERM FOLLOW-UP**

Over the ensuing 5 years, CB continued to be plagued by recurrent hemoptysis, sputum production, and obstructive lung disease requiring frequent use of oral, nebulized, and intravenous antibiotics and occasional inpatient care. Despite the noted symptoms, his lung function has remained quite close to normal without further reduction in his lung flows. A stool sample was eventually tested for elastase and was found to be normal, providing strong evidence for retained pancreatic function.

**DISCUSSION**

With ongoing regular pulmonary treatment and follow-up, CB’s prognosis is thought to be excellent, but he will require careful monitoring for other organ complications of CF, such as CF-related diabetes, osteoporosis, pancreatitis, and liver disease. He is also at potentially higher risk for certain cancers (pancreatic and colon), and thus will require careful monitoring and screening for related signs and symptoms as he ages.

His CF presentation is unusual based on his age at diagnosis (compared to the mean age of diagnosis <2 years of age for all patients with CF), and his relatively isolated symptoms of hemoptysis, but careful questioning reveals several historical clues pointing toward the diagnosis, including his history of sinusitis and bronchitis, recurrent hyponatremic dehydration, and potential infertility. Adult patients with CF with mild lung disease, normal nutritional status with pancreatic sufficiency, and partial retention of CFTR activity are being increasingly appreciated in adult care, and the diagnosis must be considered in patients presenting with unexplained bronchiectasis and/or CF organ dysfunction.