CASE REPORT

An Unusual Presentation of Cystic Fibrosis in an Adult

Shoban Davé, MD, Shanon Honney, MD, John Raymond, MD, and Patrick A. Flume, MD

• **The diagnosis of cystic fibrosis (CF) generally is made within the first few years of life, although some cases will not be diagnosed until adulthood. For most patients the diagnosis is suggested by typical CF-related symptoms such as chronic respiratory infection or maldigestion. The authors describe an adult patient with newly diagnosed CF whose presenting abnormalities consisted of hypokalemia and metabolic alkalosis. These are known complications of CF but are not common presenting features that lead to the diagnosis of CF. The authors discuss their patient's presentation and review his metabolic manifestations of CF.** ***Am J Kidney Dis*** **45:E41-E44.**

INDEX WORDS: Cystic fibrosis (CF); adult; metabolic alkalosis; diagnosis; hypokalemia.

THE DIAGNOSIS of cystic fibrosis (CF) generally is made within the first few years of life. More than 60% of patients are identified within the first year after birth and 90% by age 8 years, although about 4% will not be identified until adulthood.[1] Most patients present with respiratory or digestive symptoms that lead to the diagnosis of CF, although some patients present with less common abnormalities. Metabolic alkalosis is a known manifestation of CF[2] but is not a common presenting feature that leads to the diagnosis of CF. The Cystic Fibrosis Foundation Patient Registry database suggests that about 5% of patients have electrolyte abnormalities, which may include metabolic alkalosis, at the time of diagnosis.[1] All of the described cases of patients presenting with this manifestation of CF have been children, most being infants, and there is only one mention of an adult.[3-7] We now describe an adult patient with newly diagnosed CF whose presenting abnormalities consisted of hypokalemia and metabolic alkalosis.

CASE REPORT

First Admission

In mid-July, a 36-year-old white man presented to a local hospital with complaints of generalized weakness, muscle cramping, and lightheadedness. He recently had begun working outdoors in a hot, humid climate and experienced excessive perspiration. There was a history of similar symptoms, with a more mild presentation not requiring hospitalization, when he had been subjected to similar working conditions while serving in the military. He reported anorexia, decreased frequency of bowel movements, and decreased ability to concentrate. He reported consuming large amounts of a sports drink, in excess of 1 gallon daily, with no obvious increase in urine volume. He denied use of diuretics or laxatives.

On admission, the patient had an oral temperature of 100.4°F with mild orthostatic hypotension. He was alert but fatigued and diaphoretic with clothing that was wet with perspiration. His mucous membranes were dry. The remainder of his physical examination findings were normal.

His initial laboratory values showed severe hypokalemia and both a metabolic alkalosis and increased anion gap metabolic acidosis (Table 1). He had a normal white blood cell count with a slight increase in his hemoglobin level (18.6 g/dL [186 g/L]), and his hematocrit value was 50.9%. Urinalysis findings showed a pH of 6.5 with a specific gravity of 1.027. Lactic acid was mildly elevated at 3.2 mEq/L (0.4 mmol/L), and his urine had trace ketones. Salicylates were undetectable. There was no other evidence of infection.

He was treated with intravenous normal saline and potassium replacement, and he quickly improved. His electrolytes returned to normal by the time of his discharge (day 3) and were still normal at his follow-up visit on day 12 (Table 1).

Second Admission

The patient was admitted to the hospital again 26 days after the first admission, again for severe hypokalemia and combined metabolic alkalosis and increased anion gap metabolic acidosis (Table 1). Urinalysis findings showed a pH of 6.0 and specific gravity of 1.025. Urine electrolyte analysis showed low sodium at less than 20 mEq/L (<20 mmol/L) and chloride at less than 20 mEq/L (<20 mmol/L) concentrations with a relatively high potassium concentration at 84.6 mEq/L (84 mmol/L). Serum renin was 21.81 ng/mL/h (6 ng/Lxs), and aldosterone was 33.1 ng/dL (0.92 nmol/L); these were measured with the patient in the supine position with normal values of 0.2 to 1.6 ng/mL/h (0 to 0.28 ng/Lxs) and 1 to 16 ng/dL (0.03 to 0.44 nmol/L), respectively. Fecal

From the Departments of Medicine and Pediatrics, Medical University of South Carolina, Ralph H. Johnson VA Medical Center, Charleston, SC.

Received July 6, 2004; accepted in revised form November 10, 2004.

Originally published online as doi:10.1053/j.ajkd.2004.11.009 on January 4, 2005.

Address reprint requests to Shoban Davé, MD, Medical University of South Carolina, 96 Jonathan Lucas St, 812-CSB, Charleston, SC 29425. E-mail: dave@musc.edu

0272-6386/05/4503-0025$30.00/0

doi:10.1053/j.ajkd.2004.11.009

Table 1. Pertinent Laboratory Values

	Prior Labs (Day −288)	Admit #1 (Day 0)	Outpatient (Day 12)	Admit #2 (Day 26)	Outpatient (Day 62)
Na (mEq/L)	142	133	143	130	143
K (mEq/L)	4.7	2.0	4.5	1.9	3.7
Cl (mEq/L)	102	72	105	65	101
HCO_3 (mEq/L)	30	38	26	43	27
Glucose (mg/dL)	98	97	94	133	79
BUN (mg/dL)	19	19	15	18	12
Cr (mg/dL)	1.1	1.6	1.6	1.6	1.3
Anion gap	10	23	12	22	15
pH		7.54		7.59	
PCO_2 (mm Hg)		53		53	
PO_2 (mm Hg)		51		78	
A-a gradient (mm Hg)		32.5		50	

NOTE. To convert sodium, potassium, chloride, and bicarbonate in mEq/L to mmol/L, multiply by 1; glucose in mg/dL to mmol/L, multiply by 0.05551; urea nitrogen in mg/dL to mmol/L, multiply by 0.357; creatinine in mg/dL to mmol/L, multiply by 88.4.

phenolphthalein was negative. Ethanol and methanol levels were not measured.

The patient was treated again with intravenous fluids and electrolyte replacement. This resulted in normalization of serum (Table 1) and urine electrolytes and hemoglobin (14.3 g/dL [143 g/L]) and hematocrit (40%) values. There was also reduction of supine renin at 8.5 ng/mL/h (2 ng/Lxs) and aldosterone at 2.5 ng/dL (0.07 nmol/L).

Additional history found a prior infertility evaluation that revealed azoospermia. After correction of his serum electrolytes, a pilocarpine-stimulated sweat test was performed. The initial test found a sweat chloride of 97 mmol/L (normal, <40 mmol/L) and a repeat test found a sweat chloride of 100 mmol/L. CF gene mutation analysis (Ambry Genetics, Costa Mesa, CA) identified 2 mutations including δF508 and 2789+2insA. There was no family history of CF. The patient had no sinus symptoms, and there were no polyps on evaluation. He denied cough or sputum production. Spirometry results showed a forced vital capacity of 4.17 L (72% of predicted) and a forced expiratory volume at 1 second of 3.34 L (71% of predicted), and a forced expiratory volume at 1 second/forced vital capacity of 0.8, and reduced flow at the lower lung volumes. Total lung capacity was 105% of predicted, and residual volume was 149% of predicted. Diffusing capacity was 105% of predicted. These results were consistent with mild obstruction with air trapping (he denied a history of tobacco use). High-resolution computed chest tomography was normal; specifically, there was no evidence of bronchiectasis or airway wall thickening. Sputum culture grew usual oral flora. The patient's nutritional status was one of obesity with a body mass index of 30.3. A single sample of stool reportedly had elevated fat content, but a 72-hour fecal fat study was never completed. Semen analysis showed azoospermia.

DISCUSSION

This case shows a highly unusual presentation of CF in a man. The diagnosis of CF depends on appropriate phenotypic features of CF and demonstration of abnormal function of the CF transmembrane conductance regulator (CFTR), the protein coded for by the CF gene.[8] Our patient has very mild manifestations of CF with only the clinical feature of azoospermia, or congenital bilateral absence of the vas deferens (CBAVD), and no evidence of sinus or pancreatic dysfunction. He has mild obstructive pulmonary impairment but no evidence of bronchiectasis, and no pathogens have been identified in subsequent evaluations. The abnormal sweat chloride tests are consistent with CFTR dysfunction in the sweat gland. The diagnosis of CF was confirmed in our patient by the identification of 2 CFTR gene mutations.

The reason our patient has such mild manifestations of CF may be related to the specific CFTR mutations. There is at least one other documented case with the combination of gene mutations seen in our patient,[9] and, like our patient, had features of CBAVD. However, that patient had sinusitis and a sweat chloride of 60 mEq/L. There was no mention of electrolyte abnormalities. There is mention of 1 case in an adult patient (age 24 years) that was diagnosed on the basis of recurrent episodes of hyponatremic dehydration; this patient had a genotype of δF508/R117H, the latter also known to be associated with mild disease.[10]

Mutations of CFTR are classified into one of several classes dependent on the mechanism by