Case Report

CYSTIC FIBROSIS PRESENTING WITH SEVERE DEHYDRATION IN AN OTHERWISE NORMAL INFANT

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ABSTRACT

A 4-month-old male infant was referred to our center with clinical signs and symptoms of severe dehydration without fever, diarrhea, vomiting, or respiratory symptoms. The patient had a history of two similar episodes at 1.5 and 2 months of age without any known underlying cause. In each attack the patient was hospitalized and improved with management of dehydration and electrolyte imbalance. The main abnormal laboratory findings on all admissions were: hypochloremic, hyponatremic, hypokalemic metabolic alkalosis. Sweat chloride was checked at all admissions, and was found to be highly abnormal at the last admission.

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Key Words • Cystic fibrosis • dehydration • metabolic alkalosis

Introduction

Cystic fibrosis (CF) is the commonest lethal inherited disease of Caucasian population, occurring with a frequency of 1 in 2000 live births.[1]

The most common manifestations of CF are respiratory symptoms and diarrhea with poor growth which usually begin in early life.[2-4] Other early clinical manifestations include meconium ileus, meconium plug, cholestatic liver disease, hypoproteinemia with edema and ascites.[5-10]

Dehydration with metabolic alkalosis without diarrhea or respiratory symptoms is an unusual, but dangerous presentation which can be fatal if not diagnosed and managed correctly.[11-13]

During the last 34 years (from 1966-2000), a total of 23 infants (age <12 month) with this unusual clinical manifestation and laboratory findings of CF have been reported in the literature.[11-18] Only one of these cases was diagnosed neonatally, and most of the remainder presented between 4 to 9 month of age.

Case Presentation

A 4-month-old male infant was admitted to the pediatric emergency room with a history of irritability, poor feeding lasting for a few hours and finally lethargy. There was no history of fever, diarrhea, significant vomiting or respiratory symptoms.

On general physical examination, there were signs of severe dehydration, which improved after correction of hypovolemia and electrolyte imbalance. Finally, he was

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completely well. The growth indices of the patient were within normal limits.

The main abnormal laboratory findings were: hypochloremic, hyponatremic, hypokalemic metabolic alkalosis and a sweat chloride level of 145 MEq/l. The patient had two previous episodes of similar clinical presentation and abnormal laboratory findings at the ages of 1.5 and 2 months.

There were no concomitant diarrhea, fever or respiratory problem.

On previous admissions, sweat chloride level was normal and the patient improved with appropriate management for hypovolemia and electrolyte imbalance.

The patient was apparently doing well between the attacks with no diarrhea or respiratory symptoms.

The family history was positive for death of a sibling in early infancy who also had clinical manifestations very similar to the present case, i.e., without history of diarrhea, vomiting or respiratory symptoms.

Discussion

The classic clinical manifestations of CF are mainly due to respiratory system involvement or pancreatic insufficiency which present with chronic cough, recurrent pneumonia, respiratory distress or chronic diarrhea with growth failure beginning usually in early infancy. In this case, there was no history of respiratory symptoms, diarrhea, fever or any other specific problems which could explain the repeated attacks of dehydration and metabolic alkalosis. Growth failure is a common finding in patients with CF. In our case, the growth indices (weight, length and head circumference) were within normal range. The patient was apparently doing well with no diarrhea or respiratory symptoms between attacks. Family history was positive for the same clinical presentation without diarrhea or respiratory symptoms which led to demise in early infancy of one of the previous siblings. This kind of unusual presentation of CF (dehydration with metabolic alkalosis without diarrhea or respiratory symptoms) seems to be the manifestation of certain genetic mutations in CF.[15-17]

Although there was no history of diarrhea, respiratory symptoms or growth failure in our patient, given the clinical manifestation, laboratory changes, and also positive family history, CF was considered as the main possibility in differential diagnosis from early admission. The major mechanisms for dehydration and electrolyte imbalance in patients with CF are: 1) diarrhea, 2) increased insensible water loss due to: a) respiratory distress with tachypnea b) excessive sweating. In our case there was no history of significant diarrhea or repiratory distress with tachypnea, that could explain his dehydration and electrolyte disturbance, therefore excessive fluid and electrolyte loss from the skin was the only possible mechanism. Considering the age distribution of reported cases, it is probable that our patient is the second youngest reported with this unusual manifestation of CF to date.

While sweat test was done on all admissions, it was non-confirmatory on the first two admissions, though highly abnormal on the last admission. It is concluded that dehydration with metabolic alkalosis without diarrhea or respiratory symptoms can be the early manifestation of CF and sweat chloride test may be misleading in early infancy.

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