Vomiting may be the Only Sign of Cystic Fibrosis: A Case Report

Kusma Kistik Fibrozis Hastalığının Tek Belirtisi Olabilir: Olgu Sunumu

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ABSTRACT

Cystic fibrosis has variable clinical presentations at different ages. We present here a-7-month-old boy admitted with vomiting, hypokalemia, hyponatremia, hypochloremia and metabolic alkalosis that corresponded to a pseudo-Bartter syndrome. He had been hospitalized twice because of vomiting of unknown origin. A sweat test had already been performed and the result was normal. He had been admitted to our pediatrics unit for diagnostic work-up. Reevaluation of physical examination, serum electrolytes, blood gases and cystic fibrosis mutation analysis finally led to a diagnosis of cystic fibrosis. We planned to call the attention to the often disregarded message that vomiting may be the only sign of cystic fibrosis. Sometimes invasive tests planned to investigate the etiology of vomiting should be postponed.

Key Words: Cystic fibrosis, hypokalemic hypochloremic alkalosis, Pseudo-Bartter syndrome

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ÖZET

Kistik fibrozis her yaşta farklı klinik bulgular ile karşımıza çıkabilen bir hastalıktır. Klinik değişkenliği zaman zaman tanı sürecini uzatabilir. Burada 7 aylık kusma ile başvuran ve hiponatremi, hipokalemi, hipokloremi ve metabolic alkoloz ile psödo-bartter sendromu tanısı konulan bir kistik fibrozis hastası sunulmaktadır. Hasta daha önce iki kez kusma etyolojisi araştırılmak için dış merkezlerde tetkik edilmiştir. Hastaya yapılan ter testi normal bulunmuştur. Hastaya yapılan ileri değerlendirme ile, kistik fibrozis gen analizi ile tanı konulmuştur. Kusma, kistik fibrozis hastalığının tek belirtisi olabilir. Hastalara kusmaya yönelik ileri invazif değerlendirmeler yapmadan once akılda tutulmalıdır.

Anahtar Sözcükler: Kistik fibrozis, hipokalemik hipokloremik metabolic alkaloz, psödo-Bartter sendromu

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INTRODUCTION

Vomiting should be accompanying many situations in infants and needs to be evaluated carefully. Vomiting can be arisen from anatomic disorders, electrolyte imbalances, infections and may be even physiological in infants. Pseudo-Bartter's (PB) syndrome may be one of the reasons of vomiting, which mimics the manifestations of Bartter's syndrome, can be caused by hyponatremic, hypochloremic dehydration with metabolic alkalosis secondary to vomiting. Other possible reasons are diarrhea (chloride losing diarrhea), hypertrophic pyloric stenosis, perspiration, drug (diuretic) abuse and cystic fibrosis[1]. We present here a-7-month-old boy with vomiting diagnosed with cystic fibrosis to call the attention to the often disregarded message that vomiting may be the only sign of cystic fibrosis suggesting the PB syndrome.

CASE REPORT

A-7-month old male infant was admitted with poor feeding, vomiting and lethargy. He had no diarrhea. He was born at full-term gestation after an uncomplicated pregnancy with a birth weight of 3500 gr. He had no congenital abnormalities and his family history was unremarkable. The parents were not relatives. In his past medical history, he had been hospitalized at the age of 3 and 5 months because of vomiting. He had been evaluated for the etiology of vomiting but the laboratory evaluation revealed no pathology and he had been followed-up outpatiently. Physical examination revealed signs of dehydration. His blood pressure was normal. Laboratory examination revealed hyponatremia (128 mEq/L), hypochloremia (61 mEq/L), hypokalemia (2.89 mEq/L), and metabolic alkalosis (blood pH 7.5, bicarbonate 53 mmol/L).

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Urine electrolytes and urine microscopy were normal. Urinary ultrasound revealed increased renalparenchymalthicknessinboth kidneys. He was treated with intravenous fluids and sodium and potassium supplementation which resulted in the normalization of his serum electrolytes. The diagnosis of cystic fibrosis (CF) was arisen and sweat test was performed. Sweat test was 40 mEq/l by Gibson-Cooke method. The sweat test was repeated and concluded in the same way. So CF mutation analysis was performed and detected D110H (c.328 G>C) and (delta)508(c.1521_1523 del CTT compound heterozygosis and CF was diagnosed. He remained stable during 6 months of follow-up.

DISCUSSION

Here we present a patient with a diagnosis of CF despite the pre-established and normal examination for vomiting. PB syndrome was diagnosed through these clinical and laboratory tests.PB syndrome, which mimics the manifestations of Bartter's syndrome, can be caused by hyponatremic, hypochloremic dehydration with metabolic alkalosis secondary to vomiting, diarrhea (chloride losing diarrhea), hypertrophic pyloric stenosis, perspiration, drug (diuretic) abuse and so on [1]. Persistent watery diarrhea with a high concentration of chloride in stool is the key finding in the differentiation of congenital chloride diarrhea. Our patient had no diarrhea and didn't use any drugs and abdominal ultrasound excluded hypertrophic pyloric stenosis. Infants with cystic fibrosis (CF) are also prone to develop PB syndrome[2]. The diagnosis of cystic fibrosis was arisen and sweat test was performed. Sweat test was 40 mEq/l by Gibson-Cooke method. The sweat test was repeated and concluded in the same way. There are two ways of performing sweat test in children. Sweat test can be done by Gibson Cooke and Macroduct collection method. Both methods use iontophoresis followed by sweat collection. In the Gibson Cooke method, the collected sweat is assayed by titration. In the Macroduct collection method, sweat chloride analysis is measured by conductivity. In sweat test with Gibson Cooke method concentration of chloride in the range of 0-40mmol / L is accepted as normal, between 40 and 60 mmol / L levels are accepted as suspicious, and 60 mmol / L or more is interpreted as high. In the Macroduct collection method, the conductivity value found in the range of 0-60 mmol / L is accepted as normal, 60-90 mmol / L are accepted as suspicious, and 90 mmol / L and above are interpreted as high (3-6). In the results of suspicious ranges it is suggested to reperform the test. But the definite diagnosis depends on the genetic analysis.

So CF mutation analysis was performed to prove the diagnosis in our patient. CF mutation analysis detected D110H (c.328 G>C) and (delta)508(c.1521_1523 del CTT compound heterozygosis and CF was diagnosed. Mutations in Turkish CF patients are heterogeneous. The most common one is Delta F508 mutation[3]. CF may not present with typical pulmonary or gastrointestinal signs as in our child. Pseudo-Bartter syndrome as an initial presentation of cystic fibrosiswas detected to be 12% in one study[2], 16.5%[4] and 46%[1] in other studies. The incidence of PB was higher in cases that were diagnosed during one year of age [2,5,6].

When a patient has a PB clinic, even sweat test is normal, CF has to be ruled out especially by mutation analysis. Vomiting should be the only sign of the patients.

CONCLUSION

Vomiting may be the only marker of cystic fibrosis suggesting the metabolic decompensation accompanying PB syndrome. Sometimes invasive tests planned to investigate the etiology of vomiting should be postponed.

Conflict of interest

No conflict of interest was declared by the authors.

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