J Surg Med. 2019;3(1):91-93.
DOI: 10.28982/josam.470193

Case report
Olgu sunumu

Journal of Surgery and Medicine

e-ISSN: 2602-2079

The unexpected surgical emergency in a child with cystic fibrosis: An acute appendicitis with unusual presentation

Kistik fibrozlu bir çocukta beklenmedik cerrahi acil: Atipik sunumlu akut apandisit

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Informed Consent: The author stated that the written consent was obtained from the patient presented in the study.
Hasta Onamı: Yazar çalışmada sunulan hastadan yazılı onam alındığını ifade etmiştir.

Conflict of Interest: No conflict of interest was declared by the authors.
Çıkar Çatışması: Yazarlar çıkar çatışması bildirmemişlerdir.

Financial Disclosure: The authors declared that this study has received no financial support.
Finansal Destek: Yazarlar bu çalışma için finansal destek almadıklarını beyan etmişlerdir.

Received / Geliş tarihi: 13.10.2018
Accepted / Kabul tarihi: 08.11.2018
Published / Yayın tarihi: 06.12.2018

Abstract

Cystic fibrosis is an autosomal recessive disease characterized by structural and metabolic genetic dysfunction of the exocrine glands. Abdominal pain is the most common symptom in patients with cystic fibrosis within gastrointestinal symptoms. It is more commonly related to distal ileal obstruction syndrome. In patients with cystic fibrosis, the appendix is frequently distended due to an intense eosinophilic secretion but ironically, the incidence of acute appendicitis has been reported under the general population. Acute appendicitis in patients with cystic fibrosis commonly present with atypical symptoms. We present the case of cystic fibrosis with an acute appendicitis which had atypical radiologic and laboratory findings that were not compatible with the diagnosis. The diagnosis was made on the basis of clinical examination. As a result, it is of great importance to know that acute appendicitis in patients with cystic fibrosis can present with unusual clinical and radiological findings in order to avoid delayed diagnosis and related serious complications.

Keywords: Cystic fibrosis, Appendicitis, Pediatric age group

Öz

Kistik fibrozis ekzokrin glandların yapısal ve metabolik genetik disfonksiyonu ile karakterize otozomal resesif bir hastalıktır. Kistik fibrozisli hastalarda gastrointestinal semptomlar içerisinde ağrı en yaygın semptomdur ve genellikle distal ileal obstrüksiyon sendromuna bağlıdır. Kistik fibrozisli hastalarda apendiks yoğun bir eozinofilik sekresyondan dolayı sıklıkla distandüdür ancak ironik olarak akut apandisit insidansı genel popülasyondan daha düşüktür. Akut apandisit, kistik fibrozisli hastalarda sıklıkla atipik semptomlarla kendini gösterir. Bu yazıda atipik radyolojik bulguları olan kistik fibrozisli bir akut apandisit olgusu sunuldu ve laboratuvar bulguları, tanı ile uyumsuzdu. Hastada sadece defans ve rebound gibi klasik muayene bulguları mevcuttu ve bu temelde hasta tanı aldı. Sonuç olarak, geç tanıya bağlı ciddi komplikasyonları önlemek için, akut apandisitli kistik fibrozis hastalarının atipik klinik ve radyolojik bulgularla başvurabileceği akılda tutulmalıdır.

Anahtar kelimeler: Kistik fibrozis, Apandisit, Pediatrik yaş grubu

Introduction

Cystic fibrosis (CF) is an autosomal recessive disease characterized by structural and metabolic genetic dysfunction of the exocrine glands. It is more common in the white population. Defects in the cystic fibrosis transmembrane regulator protein (CFRT) gene lead to impaired chlorine transportation of the epithelial cell in plasma membrane of organs such as the lung, pancreas, intestine, liver, epididymis and sweat glands. Lungs are the most affected organ [1]. When gastrointestinal system is affected, abdominal pain is the most common symptom in patients with CF and is frequently due to distal ileal obstruction syndrome (DIOS). Other conditions which are leading to lower abdominal pain in these patients are intussusception, volvulus, pancreatitis and fecaloma [2]. Acute appendicitis is a rare entity in CF [3]. This paper reports, with clinical and radiological findings, the case of a patient in the pediatric age group with acute appendicitis complicating CF.

How to cite / Atıf için: Doğan E, Süzen A, Doğan MM, Çelik SY, Çullu N. The unexpected surgical emergency in a child with cystic fibrosis: An acute appendicitis with unusual presentation. J Surg Med. 2019;3(1):91-93.

Case presentation

A 13-year-old patient was admitted to the public state hospital for abdominal pain and nausea. Simultaneously, chest and plain abdominal X-ray had been taken. Bilateral hilar and perihilar overload in chest X-ray were found. White blood cell (WBC) and biochemical tests were normal. There were insignificant several scattered gas shadows in the abdominal X-ray graphic and a levelling gas corresponding to fundus anatomical localization (Figure 1).

Defense and rebound were found during clinical examination, thus ultrasonography was requested. Ultrasonography revealed a distended appendix with loss of response to compression. No evidence of perforation with issue of fluid and free gas was observed. Patient was referred to university hospital for evaluation by a pediatric surgeon with an initial diagnosis of acute appendicitis.

During the interrogation, the notion of cystic fibrosis was found in the antecedents, diagnosed at the age of 2 months, followed regularly and treated with pancreatic enzymes, Proton Pump Inhibitor (PIP), vitamins and osteoporosis' medications.

Our findings were similar to those of the examinations made previously. Physical examination revealed a rebound and defense in the lower right quadrant. Hemogram and biochemistry tests were repeated and results were normal (WBC: 7600 / mm3, Hb: 15 g / dL, CRP: 1.3 mg / L). Fundus shadows were similar to those observed in the old abdominal radiography however in the medial part gas clarity of transverse colon had appeared. There was no fluid and air level but DIOS was suspected for the increase in gas shadows, the transverse colon shadow and the fact that WBC was low. Therefore, we decided to repeat the patient's ultrasonography.

The abdominal ultrasonography performed in our hospital showed an increased appendix diameter (6.5 mm) and a partial response to compression. Liquid, lymphadenopathy, perforation and abscess were not observed. In addition, the liver was granular and heterogeneous which raised the confusing question if there was a chronic liver parenchymal disease. Furthermore, splenic cranio-caudal dimension was 132 mm which was compatible with splenomegaly.

Finally, the decision of the operation was made on the basis of clinical and radiological findings. Appendectomy was done and no complications were observed. Patient was discharged on the 2nd day. Before preparing case report, the consent form was signed by family in 10.8.2018

Histopathology

The material of appendectomy consisted on 6 cm long, 1 cm wide and cross-section lumen-shaped tissue. The material was sampled into tissue cassettes after 10% formalin fixation. Following routine tissue formation, paraffin blocks were formed. 5 µm thick sections were taken from the blocks and stained with hematoxylin-eosin. The slides were examined by light microscope.

In the pathological sections; mucosa of the appendix was characterized by ulceration, lymphoid follicles (Figure 2) and acute inflammation (Figure 3) involving a large number of polynuclear leukocytes extended to the muscularis mucosa.

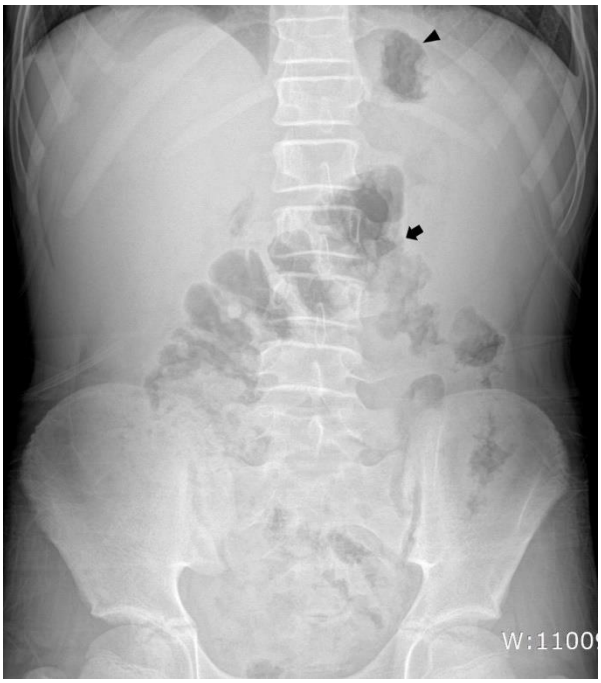

Figure 1: Arrow Head: Fundus gas, Short arrow: Increased gas shadows

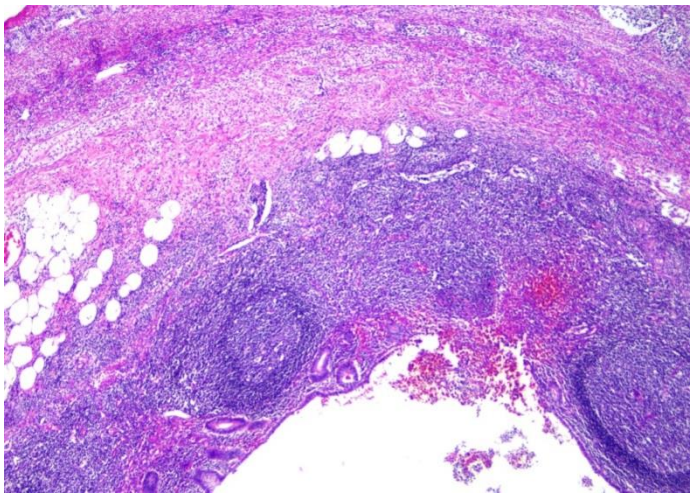

Figure 2: Ulceration and lymphoid follicles in the appendix mucosa. Hematoxylin-eosin (HE) x 40

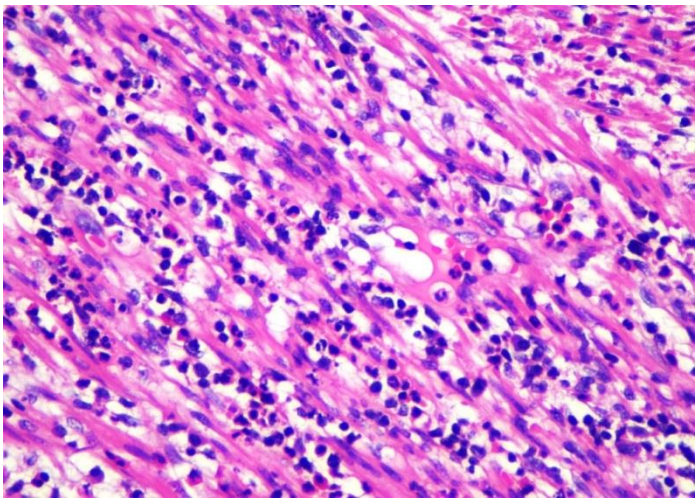

Figure 3: Acute inflammation involving a large number of polynuclear leukocytes in muscularis mucosa. Hematoxylin-eosin (HE) x 400

Discussion

The incidence of CF in Turkey is unknown. Given the prevalence in neighboring countries in Europe and the Mediterranean, it is thought to be about 20 000 CF patients in Turkey. The number of patients monitored at various centers in

Turkey is around 750-1000 [4]. The surplus of rare mutations increases the number of CF patients who are likely to come with atypical presentations [4,5]. It seems that more than 1000 mutations were associated with CF where localized in the long arm of the 7th chromosome in the CFTR gene that was found in 1989 [6]. Kilinc et al. [7] conducted a study of 83 CF patients, to determine CFTR mutations in our region (Aegean region). 36 different CFTR mutations were detected and these results indicate that Turkish population has the highest genetic heterogeneity among the studies reported so far.

In patients with CF, the appendix is frequently distended due to an intense eosinophilic secretion but ironically, the incidence of acute appendicitis has been reported under the general population [2]. In patients with CF, the incidence of appendicitis varies between 1 and 2% while in normal population rates are defined as 7% [5]. Authors attributed the low incidence of appendicitis to the protective effect of mucous secretion in the appendix in these patients. This theory has been described as a continuous distension of the appendix lumen, reducing the risk of acute appendicitis and luminal occlusion. Thus, acute appendicitis may be atypical. Symptoms may be confusing with DIOS and intussusception or the results may be masked by a prescribed antibiotic therapy to prevent pulmonary exacerbation [2]. Coughlin et al. reported 60% of the classical symptoms of appendicitis in the general population against only 45% in CF. As a result, perforation and abscess formation are higher in CF patients with acute appendicitis [8].

As mentioned above, patients with CF usually have an increased appendix diameter (>6 mm) due to an intense secretion without appendicitis. Therefore, the increase in appendix size is not a reliable finding in these patients [5]. Differentiating acute appendicitis from chronic distention of the appendix in CF patients is important for avoiding unnecessary appendectomy yet can be challenging [3,5]. It is important that the diagnosis should be supported by secondary signs of inflammation including tenderness, rebound and defense [9]. In our case, atypical radiology findings were blurred and the laboratory findings didn’t support the diagnosis. Therefore, the diagnosis was only made on the basis of clinical examination.

Conclusion

Distal ileal obstruction syndrome remains the most common cause of acute abdominal pain in CF patients and it could also mimic the symptoms of acute appendicitis. Turkey has a broad genetic spectrum for CF; therefore, we can more frequently face atypical expressions of acute appendicitis. The diagnosis of acute appendicitis and its complications in CF patients is difficult, despite the availability of modern laboratory and radiological techniques. It is of great importance to evaluate the clinical, laboratory and radiological findings together in CF patients with acute abdominal pain.

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