A Rare Cause of Severe Abdominal Pain in Children: Hereditary Angioedema

Çocuklarda Şiddetli Karın Ağrısının Nadir Bir Nedeni: Herediter Anjioödem

Sefa Sağ[1], Mustafa İmamoğlu[2], Murat Çakır[3], Elif Sağ[3]

[1] Health Sciences University, Kanuni Training and Research Hospital, Department of Pediatric Surgery, Trabzon, Turkey
[2] Karadeniz Technical University, Faculty of Medicine, Department of Pediatric Surgery, Trabzon, Turkey
[3] Karadeniz Technical University, Faculty of Medicine, Department of Pediatric Gastroenterology, Hepatology and Nutrition, Trabzon, Turkey

ABSTRACT

Hereditary angioedema (HA) is an autosomal dominantly inherited disease characterized by recurrent angioedema attacks. Angioedema is frequently seen in the arms and legs, the neck, the airways, in the genital region and in visceral organs. Edema of the intestinal mucosa can lead to transient obstruction and severe abdominal pain that can mimic acute abdomen. This can result undergoing unnecessary surgery in the patients. We presented a 15 years old girl that admitted to the emergency department due to sudden onset colic-type abdominal pain. The patient's mother had previously under gone surgery three times with the same findings who has been followed up HA. We report this case to emphasize that patients with HA may present to emergency departments with severe abdominal pain related to intestinal involvement and with findings of acute abdomen.

Key Words: Children, isolated intestinal angioedema, unnecessary surgery.

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ÖZET

Herediteranjioödem (HA); tekrarlayan anjioödem atakları ile seyreden, otozomal dominant kalıtılan bir hastalıktır. Anjioödem sıklıkla kol ve bacaklarda, yüzde, boyunda, solunum yollarında, genital bölgede ve visseral organlarda görülür. İntestinal mukoza ödemi geçici obstrüksiyona ve akut batın ile karışabilen şiddetli karın ağrılarına neden olabilir. Bu durum hastaların gereksiz cerrahi işlem geçirmesiyle sonuçlanabilmektedir. Biz, acil servise ani gelişen kolik tipte karın ağrısıyla başvuran 15 yaşındaki bir hastayı sunuyoruz. Hastanın annesi de üç kez aynı bulgular nedeniyle gereksiz cerrahi işlem geçirmişti. Biz; HA'lı hastaların, intestinal tutuluma bağlı şiddetli karın ağrısı ve akut karın bulguları ile acil servislere başvurabileceğini vurgulamak amacıyla bu olguyu sunuyoruz.

Anahtar Sözcükler: Çocuk, izole intestinal anjioödem, gereksiz cerrahi

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INTRODUCTION

Hereditary angioedema (HA) is an autosomal dominantly inherited disease characterized by life-threatening angioedema attacks. The fundamental problem in the disease is decreased synthesis of functional C1 inhibitor (C1-INH) antigenic protein in association with C1 inhibitor gene mutation. Two types have generally been described. Type 1 involves decreased C1-INH antigenic protein and decreased activity, while type 2 is characterized by normal or increasedC1-INH protein and decreased C1-INH function. In type 3, which may also be described as a subgroup, theC1-INH gene is normal. Angioedema is most frequently seen in the arms and legs, the neck, the airways, in the genital region and in visceral organs. Edema of the intestinal mucosa can lead to transient obstruction and severe abdominal pain that can mimic acute abdomen (1). Although intestinal angioedema occurs under the influence of genetic factors, it can also be triggered after various medical treatments (ACE inhibitors)(2). Some patients may undergo unnecessary appendectomy or explorative laparoscopy before HA is diagnosed(3).

We report this case in order to emphasize that patients with HA may present to emergency departments with severe abdominal pain related to intestinal involvement and with findings of acute abdomen.

CASE REPORT

A 15-year-old girl who has been followed up HA type 1 for seven years [C4 level 5.4 mg/dl (10-40 mg/dl), C1-INH level 6.1 mg/dl (24-40 mg/dl)]admitted to the emergency department due to sudden onset colic-type abdominal pain. The patient's mother had previously undergone surgery three times with the same findings who has been followed up HA.The vital signs of the patient were normal and physical examination revealed no edema in the extremities and oral mucosa, while diffuse tenderness and defense were in the abdomen at this time. At laboratory examination, blood count, biochemistry, acute phase reactants, amylase and complete urine test were normal. Computerized abdominal tomography (CAT) revealed wall thickening of up to 11 mm in the duodenum and proximal jejunum and free fluid in the abdomen (Figures 1, 2).There were no signs of appendicitis and morphology of liver, gallbladder, kidneys were normal. On the basis of these findings, intestinal angioedema was considered in this patient with HA, and fresh frozen plasma as a dose of10 ml/kg was administered. The patient's abdominal pain improved dramatically and the findings of acute abdomen resolved entirely.

Address for Correspondence / Yazışma Adresi: Sefa Sağ, MD, Health Sciences University, Kanuni Training and Research Hospital, Department of Pediatric Surgery, Trabzon, Turkey E-mail: drsefa51@mynet.com

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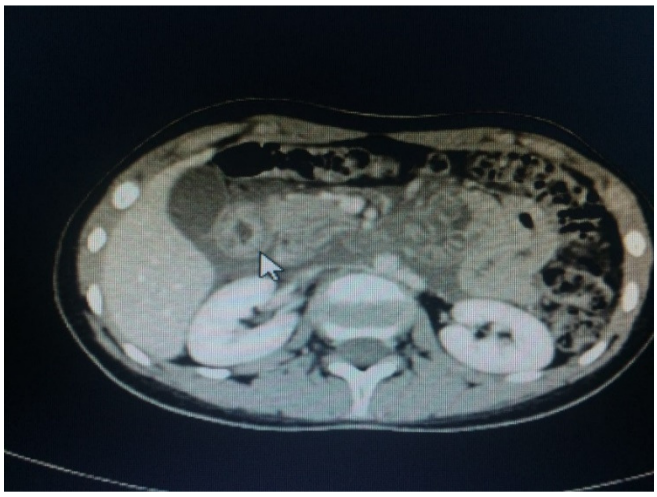

Figure 1: Duodenal wall thickening

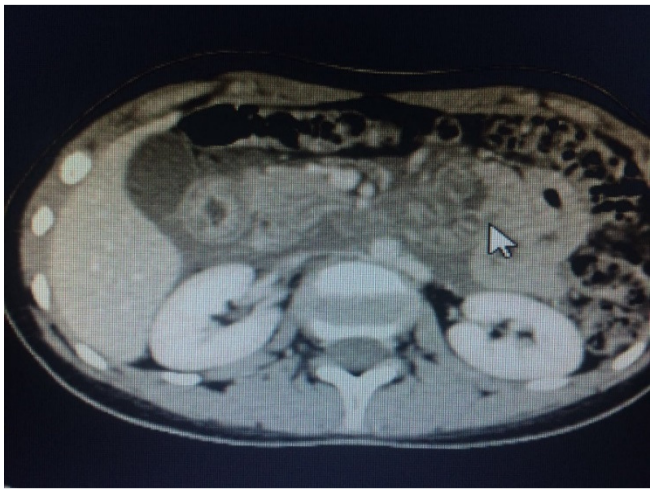

Figure 2: Jejunal wall thickening and free fluid in abdomen

DISCUSSION

Hereditary angioedema is an autosomal dominantly inherited disease first described by William Osler in 1888.The edema that occurs during attacks is the result of the vasodilator effect of bradykinin accumulation (2). The prevalence of HA is 1/50.000. There isn't difference in prevalence between sex or ethnic groups(4). Clinically, angioedema is most commonly seen in the extremities and oropharynx, but the visceral organs may also be involved. Our patient presented with severe abdominal pain and acute abdomen findings, but no cutaneous or oral mucosa edema was present.

Attacks generally begin in the first 24 hour, and the skin and digestive system are affected. Abdominal attacks proceed with severe pain, vomiting and patients may undergo unnecessary surgical procedures(5). Indeed, the mother of our patient, who was diagnosed with HA, had previously undergone three surgical operations.

Intestinal angioedema may be mistakenly diagnosed as Crohn disease, familial mediterranean fever and gastroenteritis(6).Correct diagnosis in some cases may take up to 19 years, as in cases from Japan (7).

Accurate diagnosis will become easier as the use and quality of imaging techniques such as ultrasound and tomography and unnecessary surgery will to a large extent be prevented. The bowel wall thickening on CAT may be associated with inflammatory bowel disease, local edema in the intestinal wall, hemorrhage, infectious colitis, graft-versus-host disease, and intestinal bleeding or a variant of normal (8). Indeed, wall thickening of up to 11 mm in the duodenum and proximal jejunum and free fluid in the abdomen were determined at CAT in our case. But there were no signs of appendicitis, any obstrictic patology and morphology of liver, gallbladder, kidneys were normal. Additionally laboratory findings as blood count and acute phase reactants were normal. So we thought intestinal angioedema because of clinical presentation, CAT findings and history of HA in our patient.

Recombinant human C1-INH,subcutaneousicatibant or subcutaneous ecallantide are recommended in the treatment of attacks. However, these new drugs are very expensive and cannot be actively employed in many centers. Fresh frozen plasma is generally employed during attacks(9).In this case, we treated the patient with fresh frozen plasma too and symptoms improved dramatically.

Patient and family history and well anamnesis are important to accurate diagnosis. If her history had not been well investigated, she might have undergone unnecessary surgery as other cases in the literature.

In conclusion, HA must be considered at differential diagnosis of severe abdominal pains presenting with a clinical finding of acute abdomen. Accurate investigation of the patient and family histories is important in terms of preventing unnecessary surgical interventions.

Conflict of interest

No conflict of interest was declared by the authors.

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