Hereditary angio-edema involving the gastrointestinal tract: CT findings

Abstract  We report a case of hereditary angio-edema in a young man presenting with recurrent abdominal pain for many years. The diagnosis was suspected on the basis of abdominal CT performed during an abdominal attack and was then confirmed by the measurement of serum concentration of C1 esterase inhibitor (C1-INH). To our knowledge, this is the first case reported of the hereditary form of angio-edema with isolated abdominal pain and in which the diagnosis was suggested by abdominal CT findings.

Key words  Angioneurotic edema · CT · Gastrointestinal diseases

Introduction
Hereditary angio-edema (HAE) is due to a lack of serum inhibitor of the first component of human complement (C1-INH). The uncontrolled auto-activation of C1 finally results in edema of the subcutaneous tissues, gastrointestinal tract, and the upper airway [1]. Acute abdominal pain is common for HAE and can mimic surgical emergencies particularly in patients with no other symptom. Patients in whom the diagnosis is missed may undergo appendectomy or exploratory laparotomy during abdominal attacks. Computed tomography of the abdomen performed during the acute episode of the abdominal pain may, however, be very suggestive of this diagnosis.

We report a case of HAE in which abdominal pain was an isolated symptom while the diagnosis was suggested on the basis of the abdominal CT.

Case report
A 21-year-old man was admitted to the hospital with an acute episode of diffuse spastic abdominal pain, nausea, and vomiting. The patient mentioned a history of recurrent attacks of abdominal pains during the previous 4 years. A similar attack occurred 1 year...
before and appendectomy was performed with no inflammatory abnormalities found on pathological examination. On physical examination there was abdominal tenderness while bowel sounds were present. Blood laboratory tests revealed mild leukocytosis with no other abnormalities. On the day of admission unenhanced and enhanced helical CT (collimation of 10 mm, pitch of 1.0, 120 ml of iodinated IV contrast material) of the entire abdomen was performed. Computed tomography showed a circumferential and symmetric thickening of the duodenal and proximal jejunal wall. The bowel wall showed alternated rings of high and low density (target sign) secondary to mucosal hyperemia and submucosal edema. A small amount of free fluid was identified in the right paracolic gutter (Fig.1a). Follow-up abdominal CT performed 22 h later showed an almost complete resolution of the abnormalities (Fig 1b).

On the basis of this clinical and radiological presentation the diagnosis of HAE was suggested and confirmed by laboratory findings of decrease in both antigenic and functional measurements of C1-INH [1 < 3 mg/dl (n = 22–48 mg/dl) and 16 % (n = 80–125 %), respectively]. The plasma C3 and C4 levels were also decreased [C3 = 86 mg/dl (n = 90–180 mg/dl) and C4 = 6.1 mg/ dl (n = 10–40 mg/dl)].

**Discussion**

Hereditary angio-edema is an uncommon autosomal dominant disease that is characterized by deficient activity of the inhibitor of the activated first component of the complement (C1-INH). This leads to an inappropriate complement activation which results in angioedema. Symptoms of HAE consist of episodes of cutaneous painless, nonpruritic edema of the face and extremities, and submucosal edema of the upper airway and gastrointestinal tract. Involvement of the gastrointestinal tract causes abdominal pain, nausea, and vomiting. Episodes of bowel angio-edema are self-limited and usually resolve without complication in 12–72 h [2].

Several cases of HAE with isolated gastrointestinal signs and symptoms have been reported [3, 4]. In the series reported by Frank et al., 21 % of patients with HAE had isolated abdominal symptoms [5]. Characteristics of the abdominal attacks are not pathognomonic and they are difficult to distinguish from a surgical emergency. This misdiagnosis may lead to an unnecessary abdominal surgery as in our patient. More than 30 % of patients with HAE underwent inadequate appendectomy or exploratory laparotomy during abdominal attacks [1]; therefore, it is essential for the radiologist to be aware of this condition.

Radiological findings should suggest the diagnosis of HAE. On small bowel follow-through studies the findings consist of thickened mucosal folds in the small bowel with a “stacked coin” appearance, spiculation, “thumbprinting” of involved areas, and separation of adjacent loops of bowel as a result of mural and mesenteric thickening [6]. These findings are present only when the patient is in visceral crisis and rapidly revert to normal when the attack subsides [6]; however, in the setting of acute abdominal pain and surgical emergency, abdominal CT is the most frequently performed radiological examination. The CT appearance of intestinal angioedema has been described by Ciaccia et al. in a case of acquired C1-INH deficiency. Markedly thickened walls of the distal part of the duodenum and proximal part of the jejunum was found with a complete resolution of the lesions 1 week later [7]. The same abdominal CT findings have been reported by Kim et al. [8] in three cases of acquired intestinal angio-edema due to reaction to radiographic contrast medium, and by Nasnas et al. [9] in a case of hereditary angio-edema with cutaneous and intestinal manifestations. We report a case of hereditary angio-edema with isolated acute abdominal pain confirming the usefulness of abdominal CT for early diagnosis. The suspected diagnosis was re-

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**Fig. 1a, b** A 21-year-old man with history of recurrent abdominal attacks presented with acute diffuse abdominal pain. a Enhanced CT of the upper abdomen performed during the acute episode shows a regular thickening of the duodenal (arrows) and jejunal (arrowheads) wall and small amount of ascitis on the right para
colic gutter (asterisk). b Enhanced CT at the same level as a performed 22 h later shows an almost complete resolution of bowel wall thickening and ascitis.