ACUTE ABDOMINAL PAIN WITH A SPONTANEOUS RESOLUTION AS A MARK TO THE DIAGNOSIS OF HEREDITARY ANGIOEDEMA

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Abstract: Accurate and timely diagnostics of acute abdominal pain, a common emergency, is crucial in decreasing unnecessary surgical interventions.

We present the case of a patient, Xh. M. aged 21, transported to emergency after being wakened from sleep by severe, acute abdominal pain. The pain was non-radiating, colic, and associated with flatulence, nausea and vomiting.

The family history was negative regarding Angioedema, which decreases but does not exclude the possible appearance of hereditary Angioedema.

All laboratory and imaging findings were normal, besides the low levels of C4 complement component were 4.56 mg/dl (normal values 10–40), functional C1-esterase INH was 10.29% (normal values 80–130) C1-esterase inhibitor (protein) 4.58 mg/dl (normal values 16–33), indicating HAE typ I.

Regardless of negative medical history in the family of hereditary angioedema, de novo mutation most probably led to her being the first case in the family.

The case we have presented confirms the need to include hereditary angioedema as a differential diagnostic possibility in patients with acute abdominal pain, even more so as timely and precise diagnostics enable avoidance of unnecessary surgical interventions.

Key words: hereditary angioedema, abdominal symptoms.

Introduction

Accurate and timely diagnostics of acute abdominal pain, a common emergency, are crucial in decreasing unnecessary surgical interventions.
Case Report

We present the case of a patient, Xh.M. aged 21, transported to emergency after being waked from sleep by severe, acute abdominal pain. The pain was non-radiating, colic, and associated with flatulence, nausea and vomiting.

Upon admission, vital signs were stable and, on physical examination, a moderately distended abdominal wall was presented, as well as reduced peristaltic. Palpation initiated pain. The patient did not manifest either exanthema or enanthema.

A slow progressive decrease of pain was evident during the following 10 hours upon continuous administration of analgesics, and complete resolution occurred after 24 hours, preceded by an episode of diarrhoea.

The patient recalled 3 similar, though less intense, episodes of abdominal pain in the past. The s were 2.5 and 8 years ago. She was always awakened by pain and her appetite was diminished prior to this. These attacks spontaneously resolved after 6, 9 and 10 hours respectively.

In the past, prior to these, patient had also experienced short episodes of abdominal discomfort or pain, which she neglected due to the low intensity of the pain and the spontaneous resolution. However, each following attack was more intense and longer lasting.

The medical history of the patient was negative regarding exposition to known allergens and she had never had clinically manifested Urticaria.

There was no medical history of systemic connective tissue diseases nor of Lymphoma or other haematopoetic diseases.

The family history was negative regarding Angioedema, which decreased but did not exclude the possible appearance of hereditary Angioedema. [1, 2, 12]

Approximately 25% of all HAE cases are considered to be a result of a spontaneous mutation [5].

Results

Biochemical, immunochemical values, levels of troponine, serum amylase, D-dimers, ESR and urine were within laboratory ranges.

Upon admission, ECG s excluded acute cardiovascular disease (Fig. 1). Ultrasonographic examination showed the presence of intestinal oedema upon admission (Fig. 2) leading to a possibility of Angioedema, after 24 hours the finding was normal.
Fast resolution supported abdominal HAE, manifested as an acute, intense, transient localised oedema [1]. The average duration of symptoms varies from several hours to several days, with completely normal clinical findings in the meantime [2, 12].

Oedema can present as facial, peripheral, oropharyngolaryngeal, uro-genital or gastrointestinal [3, 4].

Hepatal parenchimal morphology was regular, without focal defects. The billiary sac, pancreas, spleen and both kidneys were normal. There were no enlarged paraaortal lymph nodes. There was clearly manifested edema of the intestinal wall with moderate intraluminal stasis. Oedema was more prominent in the jejunum, with a wall thickness of 10 mm. There was a small amount of ascites in the right fossa iliaca, as well as a somewhat more pronounced sickle of fluid subhepatally. Vesica urinaria was normal. The right ovary had a cyst with a diameter of less than 1 cm. Ultrasound finding indicated an acute episode of Angioedema.

The patient’s findings of C4 complement component were 4.56 mg/dl (normal values 10–40), functional C1 esterase INH was 10.29% (normal values 80–130) C1-estrease inhibitor (protein) 4.58 mg/dl (normal values 16–33), indicating HAE type I.
Figure 2 – US findings of gastrointestinal tract
Discussion

Angioedema is classified as
- allergic
- acquired
- hereditary
- idiopathic

Table 1

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Table 2

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The clinical presentation in our patient was a typical acute, localised occurrence of gastrointestinal oedema. Idiopathic angioedema was excluded soon after admission due to its extremely rare gastrointestinal occurrence.

The patient did not use angiotensin converting (ACE) inhibitiors, known to cause oedema of the intestinal wall [6]. Allergic angioedema is usually associated with Urticaria and/or trigger.

None of those were applicable to our patient, and all the above strongly indicated an acute episode of a gastrointestinal manifestation of hereditary angioedema.

Taking into consideration the incidence of approximately 1: 50 000, hereditary angioedema is a rare, life-threatening disease characterised by unpredictable, recurrent occurrence of episodes of oedema manifested on peripheral,
facial, oropharyngolaryngeal, urogenital and abdominal locations. The underlying cause is a mutation of the Cl-INH gene [7, 8]. Low levels of Cl-INH protein disable its normal functioning, while in type II functional activity is reduced in comparison with normal quantitative findings [9].

In acquired angioedema diminished functional activity and low values of C1q are caused by formation of anti Cl-INH antibodies, while normal values and function of C1-INH are found in type III, as well as dominant inheritance as in types I and II.

Regardless of the negative medical history in the family of hereditary angioedema, \textit{de novo} mutation most probably led to her being the first case in the family.

Hyperproduction of bradykinin and kinase II, a key factor in bradykinin degradation, is a characteristic of hereditary angioedema.

Therapy strategies include the use of recombinant or human plasma-derived C1 INH, inhibitors of plasma kallikrien and antagonists of bradykinin receptors [10, 11].

Due to the infrequent occurrence of attacks in our patient, we did not recommend prophylactic therapy.

\textbf{Conclusion}

The case we have presented confirms the need to include hereditary angioedema as a differential diagnostic possibility in patients with acute abdominal pain, even more so as timely and precise diagnostics enable avoidance of unnecessary surgical interventions.

\textbf{REFERENCES}


Резиме

АКУТНА АБДОМИНАЛНА БОЛКА СО СПОНТАНА РЕЗОЛУЦИЈА КАКО ПАТОКАЗ КОН ПОСТАВУВАЊЕ НА ДИЈАГНОЗАТА НА ОЕДЕМА ANGIO HEREDITARIUM

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Точната и навремена дијагноза на акутната абдоминална болка, честа урентна состојба, е клучна во одбегнување на непотребните хируршки интервенции.

Прикажуваме случај на пациентка Џ. М. на возраст од 21 година, транспортирана како итен пациент откако од сон ја разбудила силна, акутна абдоминална болка. Болката не ирадира, со карактер е на колика, придружена со флутуленција, гадење и повраќање.

Приложи, Од, бил. мед. науки, XXXIII/2 (2012), 85–92
Семејната историја е негативна за Angioedema, што ја намалува но не ја исключува можноста за појава на хередитарен Angioedema.

Сите лабораториски наоди беа нормални, со изключение на ниските вредности на C4 компонентата на комплемент 4.56 mg/dl (нормални вредности 10–40), функционалната С1-esterase INH беше 10.29% (нормални вредности 80–130) C1-esterase inhibitor (protein) 4.58 mg/dl (нормални вредности 16–33), што укажа на HAE тип I.

Без оглед на негативната семејна историја за хередитарен Angioedema, мошно е веројатно de novo mutation да доведе до тоа прикажаната пациентка да биде прв случај во своето семејство.

Прикажаниот случај ја потврдува потребата да се вклучи хередитарниот Angioedema како диференцијално дијагностичка можност кај појава на акутен абдомен, уште повеќе што навремена и точна дијагноза овозможува да се одбегнат непотребните хируршки интервенции.

Ключни зборови: хередитарен анггоедем, абдоминални симптоми.

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