VOLUME - 11, ISSUE - 10, OCTOBER - 2022 • PRINT ISSN No. 2277 - 8160 • DOI : 10.36106/gjra

Original Research Paper

Internal Medicine

A CASE REPORT OF HEREDITARY ANGIOEDEMA PRESENTING WITH INTESTINAL SYMPTOMS.

Dr Makka Deepika	Final year internal medicine PG, Svs Medical College
Dr M C Sai Kiriti Goud	
Dr Umesh Babu M G	Professor
Dr Kirthi	

ABSTRACT Hereditary angioedema (HAE) is the deficiency or dysfunction of C1 esterase inhibitor.It may also occur due to increased activity of factor XII / estrogen levels or through an unidentified cause. It manifests the attacks of swelling involving the skin and /or mucosa or submucosa of different organs.The attacks may be a result of specific trigger or occur spontaneously.The intestinal angioedema is clinically presented with moderate or severe pain abdomen, associated with nausea, vomiting, diarrhoea and/or ascites. The 38 year old woman presents with abdominal pain diffuse associated with nausea, vomitings, lips and eyelids swelling.During this period, investigations were done which revealed a small amount of free intraperitoneal fluid.Biochemical testing were performed which revealed C1 esterase inhibitor deficiency, decreased C4 levels. She was then diagnosed as intestinal involvement with HAE. Late diagnosis is associated with high morbidity, therefore early recognition and investigation of HAE with involvement in intestinal patients with unexplained abdominal pain is important.

KEYWORDS :

INTRODUCTION

Angioedema is a self limited, localised subcutaneous or submucosal swelling which results from extravasation of fluid into interstitial tissues. Angioedema may occur in isolation or accompanied by urticaria or as a component of anaphylaxis. They occur mainly due to type 1 hypersensitivity reactions. It is marked by dramatic swelling with more pain than pruritus and minimal erythema.

The hereditary Angioedema is a rare autosomal dominant disorder characterised by quantitative or functional deficiency of C1 inhibitor. As a result,the individual presents episodes of Edema in different organs.

HAE presents itself into three distinct types. Most of the cases (80-85%) presents with HAE type I,characterised by low levels of C1-INH. Type II presents 15-20% of cases and is related to dysfunctional C1-INH, and at normal levels. Type III with normal C1 INH is related to estrogen or factor XII mutation.There are also cases with idiopathic angioedema.

The HAE is a Misdiagnosed disease, due to rarity and for not being known by many health professionals. It's prevalence varies from 1:10,000 to 1:1,50,000.On the above, present study consisted of a case report of hereditary angioedema, where they approached etiopathogenic characteristics, clinical presentation, diagnosis and treatment.

Case Report

A 38 Year old female came with complaints of swelling of lips and face and a rash on the left arm.Initially she was treated with antihistamines and steroids and discharged after 3 days.Later she presented with diffuse abdominal pain associated with nausea and multiple episodes of vomitings. pain was not associated with fever, chills, respiratory or urinary complaints.

Her vital signs were stable on presentation. On physical examination, chest was clear on auscultation, S1 S2 were heard no murmurs.Abdomen was mildly distended with diffuse tenderness. All the diagnostic investigations were done which revealed the presence of free fluid in the peritoneal cavity.Abdominal CT scan exhibited bowel wall edema without evidence of obstruction or perforation. In this period,the laboratory tests showed leukocytosis with neutrophilia and ESR ,CRP were normal.

The patient was admitted and was treated symtomatically. In view of bowel wall edema, further evaluation was done which revealed low complement C4 levels and low levels of C1 esterase inhibitor.

Hence the patient was diagnosed to have HAE type I.

DISCUSSION

HAE is an autosomal dominant disorder due to deficiency or dysfunction of C1 esterase inhibitor leading to dysregulation of complement and kallikrein-kinin systems.

The classical presentation of HAE is recurrent episodes of severe swelling over face, lips, eyelids and intestinal tract.These attacks may be triggered by minor trauma or stress may trigger an attack,however swelling often develops in the absence of any known trigger.

Symptoms of HAE may occur at any age, but most attacks starts in the paediatric age group. In the absence of any treatment, an individual can have attacks every week with most episodes lasting for 3-4 days.The presentation of HAE mimics other cutaneous disorders such as cholinergic urticaria, solar urticaria .

This case demonstrates with gastrointestinal symptoms and cutaneous symptoms without oropharyngeal and respiratory involvement.The gastrointestinal symptoms are due to bowel wall edema and may present as colicky pain, nausea, vomiting or diarrhea. The absence of fever, peritoneal signs may distinguish from peritonitis.However in few patients due to neutrophilia, hypovolemia from fluid losses may mimic peritonitis.

Treatment with C1 inhibitor concentrates, ecallantide (kallikrein inhibitor) and Icatibant (bradykinin B2 receptor antagonist) may prevent further episodes.

CONCLUSION

It is essential to know the clinical picture and complementary tests that aid in diagnosis and treatment of hereditary angioedema,since the morbidity of the patients increases with the time of disease,altering the quality of life of the patients.

Therefore we can avoid unnecessary diagnostic and therapeutic procedures,as about one-third of patients with HAE may undergo unnecessary abdominal surgeries.

Clinicians should consider angioedema as an underlying process in cases of unexplained abdominal pain.

REFERENCES

1) Kaplan AP, Greaves MW. Angioedema. J Am Acad Dermatol. 2005;53:373–388; quiz 389-392.
2) Bork K, Staubach P, Eckardt AJ, Hardt J. Symptoms, course, and complications of abdominal attacks in hereditary angioedema due to C1 inhibitor deficiency. Am J Gastroenterol. 2006;101:619–627.
3) Nzeako UC, Frigas E, Tremaine WJ. Hereditary angioedema as a cause of transient abdominal pain. J Clin Gastroenterol. 2002;34:57–61.
4) Feller EJ, Spiro HM, Katz LA. Hereditary angioneurotic oedema: an unusual case of recurring abdominal pain. Gut. 1970;11:983–8.
5) Cicardi M, Zingale L: Clinical manifestations of HAE. In: Hereditary and acquired angioedema: Problems and progress. Proceedings of the third C1 esterase inhibitor deficiency workshop and beyond. J Allergy Clin Immunol. 2004, 114: S55-58.
6) Osler W: Hereditary angioneurotic edema. Am J Med Sci. 1888, 95: 362-367. 10.1097/00000441-188804000-00004.