Hereditary Angioedema

Muhammad Hezbullah,[1] Abdul Muktadir Shafi[2]

[1]*MBBS, FCPS (Medicine), Assistant Professor, Department of Internal Medicine, Sylhet MAG Osmani Medical College Hospital, Sylhet, Bangladesh*
[2]*MBBS, Sylhet MAG Osmani Medical College Hospital, Sylhet, Bangladesh*

Conflict of Interest: None declared

This article has been peer reviewed.

Article Submitted on: 8th November 2013

Article Accepted on: 20th February 2014

Funding Sources: None declared

Correspondence to: Dr Abdul Muktadir Shafi

Address: Sylhet MAG Osmani Medical College Hospital, Sylhet, Bangladesh

Email: abdulmuktadirshafishafi@gmail.com

Cite this article: Hezbullah M, Shafi AM. Hereditary Angioedema. J Pioneer Med Sci 2014; 4(3): 127-128

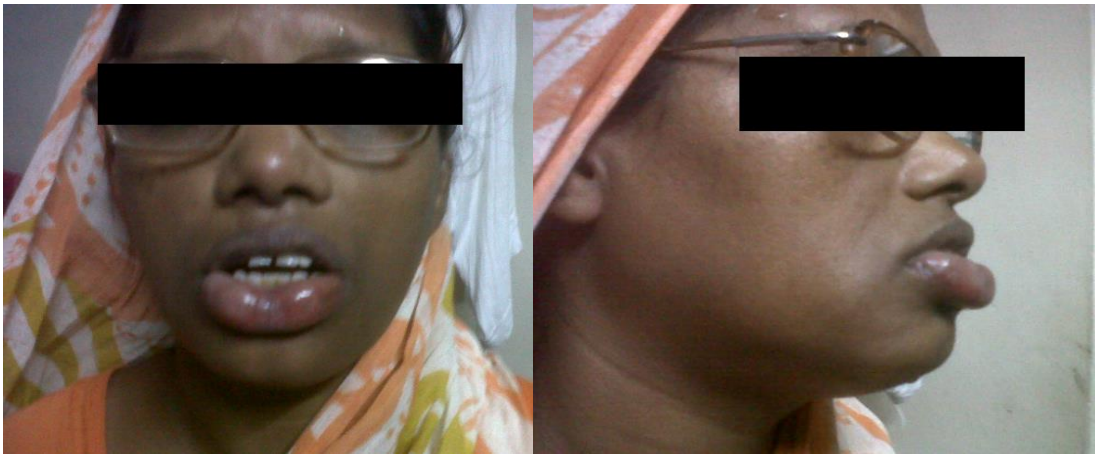
CASE REPORT

A 45 year-old woman presented to our hospital with progressive swelling of her lower lip. The patient was not complaining of shortness of breath. She had history of periodic swelling of the tongue and extremities for almost 25 years. Those episodes were spontaneous in onset without any prodromal events or any discernible cause and usually lasted from several hours to days, irrespective of treatment with antihistamines or steroids. She noted history of similar episodes in her father. She was not taking an angiotensin-converting–enzyme (ACE) inhibitor or an angiotensin II–receptor antagonist. She mentioned that the frequency of episodes increases during winter and when she is in stress. On physical examination her blood pressure was 110/80 mmHg, heart rate 110 beat/min, body temperature 36.2 ^{0}C, and respiratory rate 24/min. Diffuse edema was noted in the lower lip. There was no edema at the larynx or vocal cords. Respiratory system examination was normal. C1 esterase inhibitor levels were found to be low. The swelling worsens slowly over the first 24 hours, and then gradually subsides over the subsequent 48 to 72 hours. We diagnosed this as a case of hereditary angioedema (HAE).

DISCUSSION

HAE is an autosomal dominant disease caused by a deficiency in functional C1 inhibitor [1]. HAE is characterized by recurrent attacks of edema involving the skin, mucous membranes, gastrointestinal tract, and airway [2, 3]. Symptoms typically begin in childhood (often as early as 2 or 3 years of age), worsen around puberty, and persist throughout life, with unpredictable severity. Hereditary angioedema results from a mutation in the C1-inhibitor gene [4]. There are three types of hereditary angioedema: type I (accounting for 85% of cases), type II (15% of cases) and type III (rare). Mutations that cause hereditary angioedema type I lead to reduced levels of C1 inhibitor in the blood, while mutations that cause type II result in the production of a C1 inhibitor that functions abnormally. Unlike types I and II hereditary angioedema, type III is not caused by a deficiency of C1 inhibitor but is thought to result from the periodic accumulation of bradykinin through alternative mechanisms. Although urticaria and angioedema are common problems that affect nearly 20% of the population, HAE is a rare disorder and accounts for approximately 2% of clinical angioedema cases. HAE is suspected if a patient presents with recurrent angioedema (without urticaria), recurrent episodes of abdominal pain and vomiting, laryngeal edema and positive family history of angioedema [5]. During acute attacks, vapor-heated C1-INH(inhibitor) concentrate administration is the treatment of choice, but is

Figure 1: Hereditary Angioedema



not commercially available in our area. We managed the patient by giving fresh frozen plasma as it contains C1-INH (inhibitor). Neither corticosteroids nor antihistamines have been shown to provide a meaningful benefit during attacks of HAE. All patients with HAE who have an oro-pharyngeal attack should be closely observed in a facility where rapid intubation or tracheotomy can be performed if necessary. The history of allergy or HAE may be useful in directing therapy. Anaphylaxis should be ruled out and treated appropriately if suspected. Patients with laryngeal edema should be given epinephrine. If the laryngeal edema is secondary to mast cell activation for example anaphylaxis, it usually responds to epinephrine, whereas laryngeal edema of HAE is refractory to this therapy. For patients with HAE who have frequent or severe attacks, long-term prophylaxis with 17α-alkylated androgens or anti-fibrinolytic drugs significantly reduce the frequency of attacks. Anti-fibrinolytic drugs have therefore been recommended as the first choice in children and pregnant women who require long-term prophylaxis [6]. Patients with HAE should be advised to avoid stimuli that may precipitate attacks. Angiotensin-converting–enzyme inhibitors are contraindicated in such patients and oral contraceptive pills or hormone-replacement therapy should be cautiously used [7].

REFERENCES

1. Donaldson VH, Evans RR. A biochemical abnormality in hereditary angioneurotic edema: absence of serum inhibitor of C' 1-esterase. *Am J Med* 1963; 35:37-44
2. Eck SL, Morse JH, Janssen DA, et al. Angioedema presenting as chronic gastrointestinal symptoms. *Am J Gastroenterol* 1993; 88:436-439.
3. Winnewisser J, Rossi M, Spath P, et al. Type 1 hereditary angioedema. Variability of clinical presentation and course within two large kindreds. *J of Intern Med* 1997; 241:39-46
4. Davis AE III. C1 inhibitor and hereditary angioneurotic edema. *Annu Rev Immunol* 1988;6:595-628
5. Zingale LC, Beltrami L, Zanichelli A, et al. Angioedema without urticaria: a large clinical survey. *CMAJ*.2006; 175 (9): 1065–1070.
6. Farkas H, Varga L, Szeplaki G, Visy B, Harmat G, Bowen T. Management of hereditary angioedema in pediatric patients. *Pediatrics.* 2007; 120:e713-e722
7. Byrd JB, Adam A, Brown NJ. Angiotensin-converting enzyme inhibitor-associated angioedema. *Immunol Allergy Clin North Am* 2006; 26:725-737