CASE REPORT

A CASE OF ANGIOEDEMA: C1 INHIBITOR DEFICIENCY

Arijit Sinha[1], Suvrendu Sankar Kar[2], Subhrojyoti Mitra[3], Ujjal Bandopadhyay[4], Bikas Chandra Seth[5], Md Hamid Ali[6]

Author's Affiliations: [1]Assistant Professor, Department of General Medicine, Infectious Disease Hospital, Kolkata; [2]Assistant Professor, Department of Medicine, R.G.Kar Medical College, Kolkata; [3]Assistant Professor, Department of Medicine, Bankura Sammilani Medical College, Bankura; [4]Assistant Professor, Department of ophthalmology, Regional institute of ophthalmology, Kolkata; [5]Associate Professor, Department of Medicine, Midnapur Medical College, Midnapur; [6]Clinical Tutor, Department of Medicine, Murshidabad Medical College, Berhampur.
Correspondence: Suvrendu Sankar Kar, Email: dr_sskar@yahoo.co.in

ABSTRACT

Angioedema is rapid swelling (oedema) of subcutaneous tissue involving dermis, mucosa and sub mucosal tissues. It may be IgE dependant, bradykinin mediated, complement mediated, non immunologic or idiopathic. It may be heriditory or acquired. In our case the child was suffering from recurrent episodes of angioedema and found to be due to C1 inhibitor deficiency.

Key words: Angioedema, C1 inhibitor deficiency, complement, Urticaria.

CASE REPORT

A 13 years old Hindu male (R Sarkar) of Bankura district of West Bengal presented with recurrent swelling of face including lips and tongue, hands, genitalia and abdominal pain. Myalgia, arthalgia, itching, fever, skin rash, shortness of breath were absent. Illness was not related to any food or drugs. Episode lasts for four days and regressed with symptomatic treatment. He is the only child and no history of such illness in the family. There is no seasonal variation. He had a history of choaking sensation in the throat causing mild respiratory distress in the past.

Examination

When the child came to us during attack he was conscious but apprehensive. His face, lips, tongue were swelled and eyes were closed. There were swelling of upper part of chest, hands, feet, scrotum and penis. Urticaria, skin rash, lymphadenopathy, pallor, cyanosis, stridor, joint tenderness were absent. His pulse(106/minute),blood pressure(106/74 mm of Hg),respiration(18/minute) were normal. Examination findings of heart, lungs, abdomen and nervous system were normal.

Investigations

Patient's investigation reports were as follows: haemoglobin 12.7 gm/dl, total leucocyte count 8200 with differential count as N64, L34, E02, ESR 16 mm, bilirubin total 0.86, AST 46 IU/L, ALT 39 IU/L, urea 26mg/di, creatinine 1.1mg/di, Na 140 mEqv/L, K 4.2mEqv/L, total IgE 156 IU/ml, ANA negative, serum protein electrophoresis-normal pattern, chest x-ray normal, urine routine and 24 hours protein excretion normal, ultrasonography of whole abdomen normal. Blood for allergy profile revels allergic to egg, milk, prawn, moogdal, bringle, date, banana. Serum C1 inhibitor, protein quantification (radial immunodiffusion) level was low – 65mg/L(normal 195 to 345mg/L) with normal C1 level(28mg/dl).

Treatment and course

Patient was treated conservatively and improved. Trenexamic acid is advised for prevention of recurrence. C1 inhibitor supplementation was not used due to lack of availability.

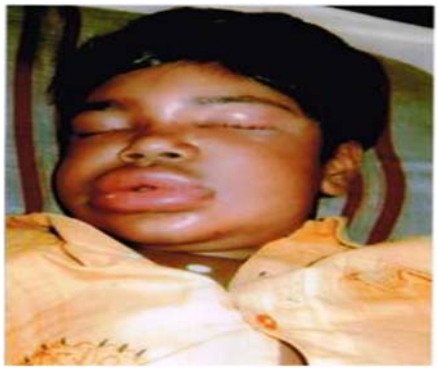

Fig 1: Photograph showing swelling of face

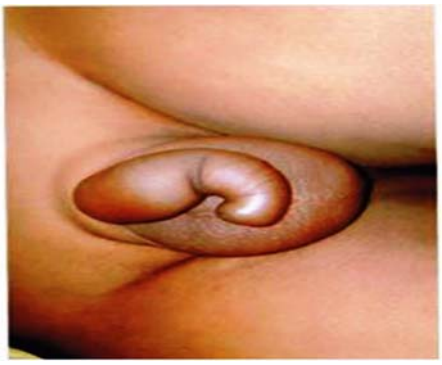

Fig 2: Photograph showing swelling of genitalia

DISCUSSION

Angioedema is swelling of subcutaneous tissue, mucous membrane and sub mucosal tissue that may affect face, extremities, genitalia, gastrointestinal tract and upper airways. It may be acquired or hereditary. Hereditary angioedema (HAE) is due to deficiency of C1 inhibitor (type 1) or abnormal function of C1 inhibitor which is mediated by bradykinin. A third type of transmitted angioedema has also been described.[1,2] Hereditary angioedema is rare genetic disorder (autosomal dominant) and 50% patients have at least one episode of laryngeal oedema in life time with incidence of abdominal pain in 70% cases.[3] It is estimated that 1 in 10000 to 1 in 50000 people is suffering from HAE and most of them presents in childhood.[4] Although most patients of HAE reports family history of angioedema, up to 25% cases have de novo C1 inhibitor deficiency.[5] Before the advent of therapy mortality in HAE was as high as 30%.[6] HAE may occur with normal C1 inhibitor concentration and function in women suggestive of X linked mode of inheritance(type 3).[7] C1 inhibitor deficiency may be acquired due to accidental consumption as a result of bindings of auto antibodies with C1 inhibitor. It is usually seen in lymphoproliferative disorder.[8]

Management of C1 inhibitor deficiency angioedema includes – acute attack management and prophylaxis. C1 inhibitor concentrate or recombinant human C1 inhibitor (rhC1 inh) are used for acute attack.[9] Kallikerin inhibitor (ecallantide) and selective bradykinin beta2 receptor antagonist (icatibant) are useful alternative.[10] Fresh frozen plasma contain C1 inh and may be used in acute attack when preferred drugs are not available.[11] Drugs used for prophylaxis are danazole, antifibrolytic agent (eg trenexamic acid). Danazole has got some serious side effects and to be monitored regularly. Antifibrinolytic agents are preferred during pregnancy. Short term prophylaxis for invasive procedure is necessary. Transfusion of C1 inhibitor 24 hours before, fresh frozen plasma on that day or Dadnazole/fibrinolytic agent 5 days before and 2 days after the procedure are preferred choice.[12,13] Patient to be vaccinated against blood product related infection.

In our case the patient was presented with recurrent episodes of angioedema without urticaria, with low C1 inh level in blood. All these suggestive of angioedema due to C1 inh deficiency type 1, but family history is lacking, the patient is now on prophylaxis with trenaxamic acid.

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