European Journal
of Case Reports in
Internal Medicine

An Unusual Case of Cerebral Oedema

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Abstract

Hereditary angioedema (HAE) is a rare genetic disorder transmitted as an autosomal dominant trait, characterized by reduced plasma concentration or by the presence of non-functional C1 esterase inhibitor. Oedema caused by HAE mostly affects the skin and bowel and can induce swelling of genitalia. Oedema can be life threatening if it causes swelling of the larynx with obstruction of the airways.

We describe the case of a 52-year-old man who presented a neurological emergency (coma), where the remarkable localization of the clinical manifestation and the unusual symptomatology hindered the correct diagnosis.

Keywords: Hereditary angioedema, coma, neurological emergency, C1 esterase inhibitor

Received: 02/06/2014

Accepted: 03/10/2014

Published: 02/12/2014

How to cite this article: Giardino F, Rizzotto A, Neri S. An Unusual Case of Cerebral Oedema. *EJCRIM 2014;***1:**doi: 10.12890/2014_000092

Conflicts of Interests: The authors declare that they have no conflicts of interest related to this research.

Case report

A 52-year-old man had headaches for two days. On the third day, he presented with transient mental confusion, dysarthria and dysmetria and came to the hospital emergency room (ER). He presented with numbness and loss of consciousness: first-degree coma was diagnosed. Haematochemical tests ruled out metabolic alterations. Glycerol was administered as an anti-oedema agent. A CT scan revealed the presence of marked cerebral oedema without space-occupying lesions or vascular involvement. Neurological symptoms worsened over the next 12 h despite the anti-oedema treatment. Coma progressed towards the second degree. The neurosurgical staff was alerted and a magnetic resonance imaging (MRI) (*Figs. 1 and 2*) scan with contrast was performed. The presence of the oedema was confirmed: vascular causes were ruled out.

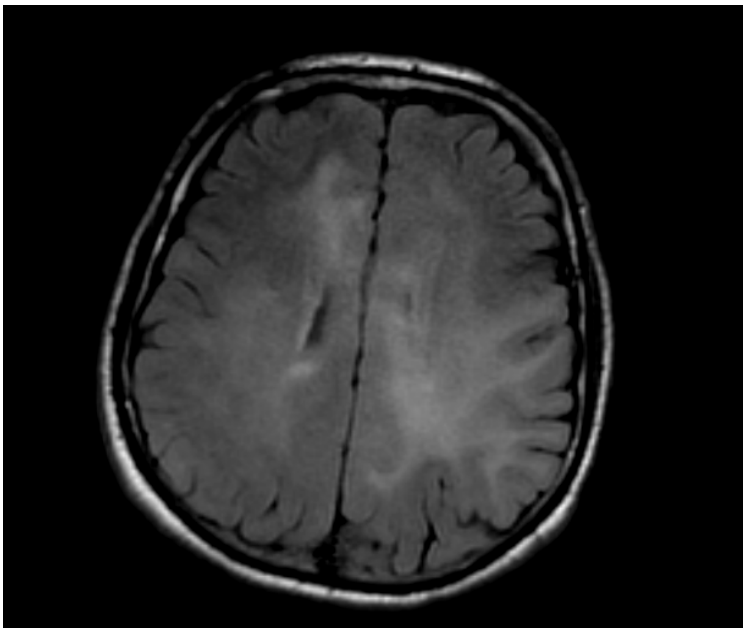

Figure 1. Contrast-enhanced MR image (T2) during the course of angioedema with partial effacement of cerebral sulci without midline shift.

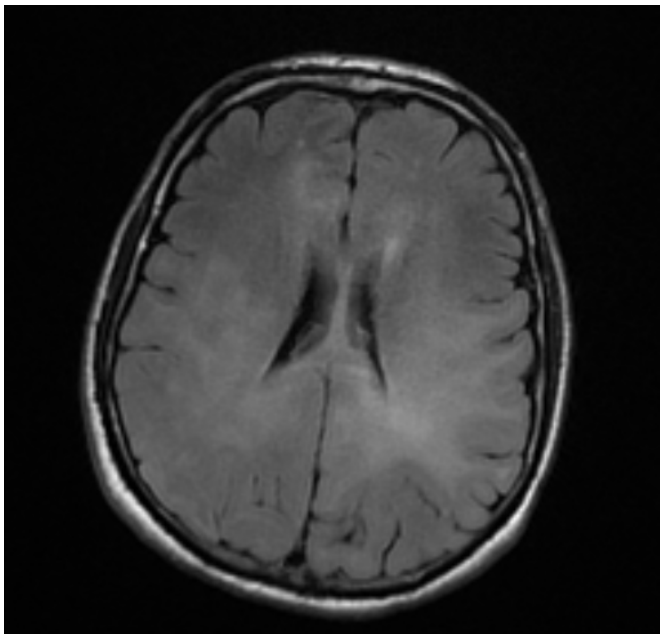

Figure 2. Contrast-enhanced MR image (T2) showing the lateral ventricles.

As the symptoms could be attributed to toxic external causes, the emergency department and poison control centre were alerted. The ER personnel gathered the information that the patient was a carrier for the genetic disorder hereditary angioedema (HAE). Prompt i.v. administration of two C1 esterase inhibitor (C1-INH) boluses (Berinert 10 ml Mix2Vial™, CSL Behring, Australia) drastically improved the patient's condition: the symptoms began to dissipate after 4 h, and he was revived from the coma after 12 h; the symptoms completely disappeared in 48 h.

Discussion

HAE is a rare disorder transmitted as an autosomal dominant trait. The disease is disabling and can be lethal. The clinical manifestations of HAE are self-limiting episodic bouts of well-circumscribed and non-itching swelling of the deep cutaneous, subcutaneous, submucosal and subepithelial tissues lasting 2–5 days. Oedema caused by HAE is dense, aching and not compressible; mostly affects the skin and bowel and can induce swelling of genitalia. Oedema can be life-threatening if it causes swelling of the larynx with obstruction of the airways [1–5].

We know three forms of this condition: Type I HAE, which is characterized by impaired synthesis and low plasma concentration of a functionally active C1-INH; Type II HAE, which is characterized by a dysfunctional C1-INH molecule that is synthesized in normal amounts (HAE is diagnosed when the C1-INH level is lower than 50% of normal); and Type III HAE, which is characterized by normal complement levels, but this can be caused by factor XII gene mutations [5].

C1-INH inhibits components of the complement, contact (factor XII and kallicrein), coagulation (factor XI and thrombin)

and fibrinolytic system and regulates the generation of vasoactive peptides, of which bradykinin is considered the most important. Current therapeutic management consists in purified C1-INH concentrate or in peripheral bradykinin inhibitor [5].

This unusual case describes a neurological emergency consisting of a comatose state caused by cerebral oedema.

Cerebral localization of angioedema is certainly unusual and it seems to have been reported only once in the literature [6]. However, the vasodilation induced by C1 inhibitor deficiency and accumulation of bradykinin can theoretically give rise to oedema in any perivascular tissue. It is also known that, generally, the oedema may occur where pressure is applied or trauma occurs. However, it supervenes very often without any apparent cause in the absence of trauma or pressure [1–5]. Brain oedema can be triggered by other unknown causes.

HAE may constitute a neurological emergency. This unusual case shows a neurological emergency consisting of a comatose state caused by cerebral oedema. Only the correct diagnosis of the disorder and specific C1 esterase inhibitor replacement therapy was able to resolve the symptoms.

In our case, correct diagnosis of the clinical picture simulating a stroke and appropriate treatment were the result of a careful history. Neither Nuclear Magnetic Resonance (NMR) nor the anti-oedema treatment facilitated diagnosis or improved outcome. In contrast, documentation of the patient's case history and pathophysiological analysis ruled out other possible underlying organic causes and led to the administration of the only therapy capable of resolving the clinical picture.

Therefore, it is always appropriate to prolong observation and to collect a more accurate history from patients or relatives in emergency cases presenting conflicting symptoms.

Finally, we recommend that patients with HAE should always carry a record of the diagnosis and emergency treatment on a document attached to their identity card.

Learning Points

- HAE can mimic neurological emergency.
- HAE is potentially fatal.
- A correct diagnosis and appropriate therapy can heal the patient quickly.

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