Mediastinal Angioedema: A Rare Manifestation of Hereditary Angioedema

M. Andreina Marques-Mejias, MD, MSc, Ana Entrala, MD, Rosario Cabañas, MD, PhD, and Teresa Caballero, MD, PhD

Hereditary angioedema due to C1-inhibitor deficiency (C1-INH-HAE) is a rare disease with variable symptoms, unpredictable course, and a challenging diagnosis.1

We present the case of a 58-year-old White male diagnosed with C1-INH-HAE who attended the emergency room (ER) because of thoracic pain.

He had had his first peripheral angioedema (AE) attack at 11 years and had been diagnosed with C1-INH-HAE 13 years later. His C1-INH-HAE diagnosis was confirmed in our unit at the age of 41 years with low C4 (7.13 mg/dL; 6-48 mg/dL), antigenic C1-inhibitor (8.46 mg/dL: 16.0-33.0 mg/dL), and functional C1-inhibitor (22.3%; >50) and a mutation in the SERPING1 gene.

He was managed with on-demand treatment of the acute AE attacks with intravenous plasma-derived C1 inhibitor concentrate (Berinert; CSL-Behring, Marburg, Germany) or subcutaneous icatibant acetate (Firazyr; Shire HGT, currently part of Takeda, Zug, Switzerland). At 53 years, there was a clear worsening of the condition, with multiple peripheral, abdominal, and upper airway angioedema episodes, mainly triggered by stress. Attenuated androgens (danazol, estanzolol) were not considered for long-term prophylaxis because of an antecedent history of hemochromatosis. The patient was offered long-term prophylaxis with intravenous plasma-derived C1 inhibitor concentrate but refused to initiate the treatment.

Age 58 years, he required assessment at the ER of another hospital because he had dysphagia, inspiratory dyspnea, and stabbing chest pain radiating to the back. Pain worsened when he was in standing position and relieved in the supine position and when leaning forward. There was no cough, peripheral angioedema, cutaneous lesions, fever, or other associated symptoms. No other potential triggers were identified.

On physical examination, he was eupneic with normal vital signs. There was no evidence of skin lesions or angioedema. The only abnormal laboratory value was an increased D-dimer of 6.52 μg/mL (0.15-0.5 μg/mL). Electrocardiogram did not show any acute or chronic alterations. Chest X-ray was performed but did not show any alterations. Because of the increased D-dimer and the pain characteristics, computed tomography (CT) angiography of the chest was performed (Figure 1A), which showed a 7 × 4 cm mediastinal soft tissue enlargement around the trachea, esophagus, principal bronchi, left atrium, and diaphragm. He was diagnosed with a bronchogenic tumor. During his stay in the ER, pain improved with analgesics. He remained stable without any associated symptoms.

Two days later a new CT was performed in a private practice setting, which showed nearly normal results, except for slight pleural edema (Figure 1B).

Taking into account the transient characteristics of the images and the increase in plasma D-dimer levels known to occur in hereditary angioedema,2 the diagnosis of bronchogenic tumor was ruled out and the final diagnosis was mediastinal AE attack. Possible mediastinal angioedema had already been described,1 but this is the first reported case of mediastinal angioedema with a complete study during the episode including CT angiography of the chest.

The key findings supporting this diagnosis include the medical history, the transitory nature of radiographic findings, and the absence of other data supporting an alternative diagnosis. We advise to perform radiographic imaging in patients with C1-INH-HAE and thoracic pain to exclude mediastinal angioedema.

REFERENCES

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FIGURE 1. (A) Findings on acute CT angiography of the chest depicting enlargement of mediastinal soft tissue around the trachea, esophagus, principal bronchi, left atrium, and diaphragm (single arrow). Small bilateral pleural effusions (double arrows). (B) Resolution of mediastinal swelling seen on follow-up CT 2 days later.