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Case report

A tell tale handshake

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ABSTRACT

Myotonic dystrophy is a group of inherited disorders called muscular dystrophies. Clinical presentation of this disease is characterised by progressive muscle weakness with myotonia, cataracts, infertility (in males) and cardiac conduction defects. We present a case of a 35 year old male with lung abscess, later diagnosed to be a case of myotonic dystrophy. Lung abscess is an uncommon presentation of this disease and has never been reported before.

1. Introduction

Myotonic Dystrophy (DM) is a clinically heterogeneous autosomal dominant disorder [1]. It is caused by an unstable trinucleotide repeat expansion containing CTG, located in the 3′ untranslated region of chromosome 19q13.3 [2]. Clinically the disease presents most commonly as weakness, wasting, myotonia or myalgia of the skeletal muscles, or sometimes affects other organs like eye, heart, brain, endocrine glands, gastro-intestinal tract, skin, skeleton and peripheral nerves [3]. Severity co-relates with the number of repeats of CTG and this disease shows anticipation i.e. the disease presents earlier and in a much more severe form in the offspring. Age of onset is usually twenties or thirties, although they can occur at any age.

2. Case

35 year-old hispanic male with history of Diabetes Mellitus and inguinal hernia presented with two-week history of productive cough and weight loss. He denied hemoptysis, fevers, chills, recent travel or sick contacts. Initial physical exam was significant for left-sided rhonchi. Chest radiograph revealed a lingular opacity with associated effusion. He was started on broad-spectrum antibiotics, and subsequent chest CT (Fig. 1) showed a left upper lobe consolidation with a large multi-locular cavitation and air-fluid levels. On our evaluation patient appeared older than stated age, with noted temporal wasting, poor dentition and gynecomastia. Additionally, subtle signs of atrophy were seen in the upper extremities with abdominal obesity and a left inguinal hernia. He answered questions appropriately but with slowed mentation and mini-mental exam was 25. Bilateral ptosis was evident with normal pupillary reflexes. Cranial nerves were intact. Motor strength was grossly 4/5 without fasciculations. When shaking his hand, a delay in relaxation of the thumb was seen, and percussion of the thenar eminence elicited a contraction of the thumb. Furthermore a delayed grip release was evident. Laboratory investigations resulted negative for HIV and Tuberculosis, however respiratory cultures grew gram-negative bacilli and gram-positive cocci in pairs. Later, a barium swallow (Fig. 2) showed laryngotracheal aspiration. Based on the described findings a clinical diagnosis of DM (Steinert's disease) was made. He was started on an eight-week course of antibiotics, improved significantly and was discharged with Neurology and Pulmonology follow-up.

3. Discussion

The prevalence of myotonic dystrophy range from 1:100,000 in some areas of Japan to 1:10,000 in Iceland, with an overall estimated worldwide prevalence of 1:20,000 [4]. Our patient presented with a large lung abscess from aspiration and hypoventilation secondary to intercostal and diaphragm myotonia.

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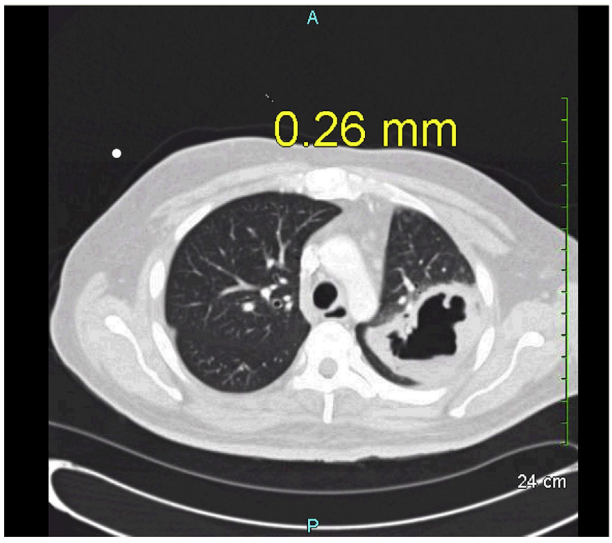

Fig. 1. CT Chest: Left upper lobe consolidation with multilocular cavitation.

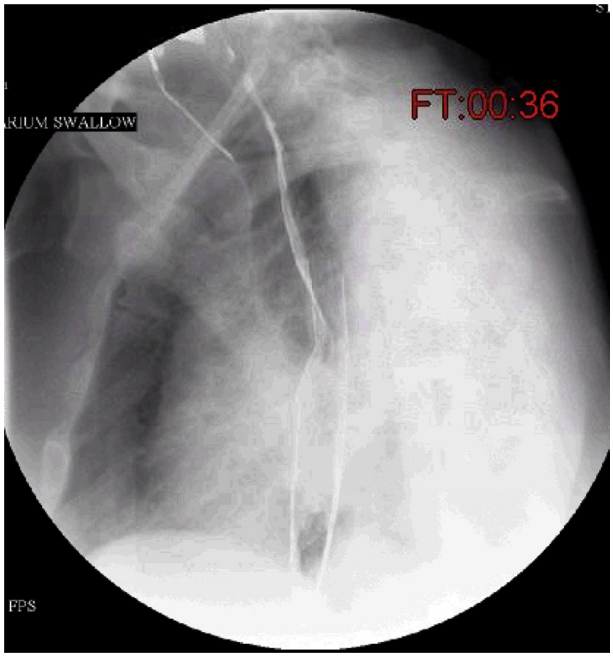

Fig. 2. Barium swallow: Neuromuscular dysfunction of the swallowing mechanism with significant laryngotracheal aspiration.

This case highlights the importance of identifying and classifying characteristic findings of DM in the setting of other conspicuous, and often confounding symptoms. In this disease, skeletal and smooth muscle involvement is widespread predisposing affected patients to chronic aspiration, pneumonia or respiratory failure, the most common cause of death [5]. Hannon et al., in 1986 reported a case of aspiration pneumonia postoperatively, in which subsequent clinical examination resulted in the diagnosis of DM [6]. Aspiration is more likely post operatively because problems like decreased ventilator capacity, depressed laryngeal reflex and prolonged muscle contraction with depolarizing muscle relaxant accumulate [7]. Muscle involvement ultimately can affect progressively the myocardium and its conduction system resulting in arrhythmia [8]. Digestive symptoms have been reported to appear up to 10 years before the musculoskeletal symptoms [9]. Diagnosis, although clinical, could be difficult in atypical or early presentation making electromyography a necessary tool. Genetic testing is advised, as this is a dominant disease.

4. Conclusion

It is imperative to perform a thorough physical exam and have a wide differential, even in patients with common presentations. Given the potentially grave consequences of aspiration pneumonia, we should consider DM 1 as an etiology to prevent recurrences.

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Author contributions

Praneet Wander, Adedapo Iluyomade, Paul Sanmartin and Akriti Gupta wrote the manuscript and reviewed literature. Mary O'Sullivan was involved in the diagnosis and management of the case and critically reviewed the manuscript and is the article guarantor.

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