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OESOPHAGEAL DYSMOTILITY IN A PATIENT WITH CHARCOT-MARIE-TOOTH DISEASE: REPORT AND LITERATURE REVIEW

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ABSTRACT

Oesophageal dysmotility is a serious condition characterised by impaired coordination of oesophageal smooth muscle contractions, which can be secondary to a variety of causes including infection, inflammation and malignancy. The presenting symptoms are variable and include chest or epigastric pain, food regurgitation, heartburn or cough, making it difficult to distinguish. Diagnostic modalities and treatment strategies vary depending on the underlying cause. Once oesophageal dysmotility is suspected, a thorough evaluation is essential as the management strategies and prognosis of the condition differ significantly based on the underlying pathology. A multidisciplinary approach and clinical expertise are essential for optimal patient care and treatment. While neuromuscular disorders are associated with swallowing dysfunction due to oropharyngeal muscle involvement, oesophageal smooth muscles involvement is rare. This case highlights the importance of careful and frequent evaluation of both respiratory and gastrointestinal smooth muscle function, particularly in patients with chronic neurological disorders.

KEYWORDS

Charcot-Marie-Tooth disease, dysphagia, smooth muscle, neurological disorder, aspiration pneumonia, oesophageal dysmotility

LEARNING POINTS

- Charcot-Marie-Tooth disease has the potential to involve smooth muscles, including those of the diaphragm and gastrointestinal tract.
- Regular and detailed evaluation of respiratory and swallowing functions is advised for patients with neurological disorders to monitor for early signs of dysfunction.
- Aspiration pneumonia should always be in the differential in patients with recurrent pneumonia and warrants thorough evaluation to ensure appropriate diagnosis and management.

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INTRODUCTION

Charcot-Marie-Tooth disease (CMT) is a common inherited neuro-degenerative condition mainly affecting the peripheral nervous system, leading to motor and sensory neuropathy as well as distal muscle atrophy. The disease is slowly progressive and may eventually involve proximal muscles. Involvement of smooth muscles, such as the diaphragm and oesophagus, is extremely rare[1]. We present a patient with advanced CMT who was admitted to the hospital with aspiration pneumonia and was found to have oesophageal dysmotility.

CASE DESCRIPTION

A 54-year-old male with a history of CMT diagnosed at the age of 3, complicated by wasting and contractures in distal muscle groups of both the upper and lower limbs, now had full dependency on a wheelchair. However, his higher mental functions were intact, and he was able to tolerate a regular diet. The patient presented with cough, fever and shortness of breath lasting three days, associated with chills and sputum production. On further questioning, the patient denied any previous history of aspiration, dysphagia or adenophagia, but mentioned experiencing occasional gastroesophageal reflux disease-like symptoms. On examination, the patient was tachypnoeic and tachycardic. He appeared to be in moderate respiratory distress and was unable to speak in full sentences. The chest examination reviewed diffuse rhonchi in all lung fields. The remainder of the examination was positive for distal muscle wasting and contractures in both upper and lower limbs. A computed tomography scan of the chest showed bilateral patchy consolidation concerning aspiration pneumonia and the oesophagus was dilated with fluid secretion *(Fig. 1)*.

The patient was started on empiric antibiotics and supportive care; he was kept 'nothing by mouth' pending swallow evaluation and further aspiration risk assessment.

Bedside swallow evaluation showed a normal pharyngeal phase of swallowing, however there remained high risk aspiration with intolerance of liquids and solids. Given the presence of oesophageal dilation and fluid stasis, the patient underwent an oesophagogastroduodenoscopy for diagnostic and potentially therapeutic purposes. Interestingly, the results were normal, showing no pathological findings. A barium oesophageal swallow test showed moderate dilatation in the upper oesophagus and severe oesophageal dysmotility. The dye did not clear through the oesophagus throughout the procedure, with no features of lower oesophageal sphincter relaxation failure *(Fig. 2)*.

The swallow team initiated a gradual diet approach with daily swallow training to advance diet as tolerated while reducing aspiration risk. At the time of discharge, the patient was able to tolerate a soft bite-sized diet with thickened liquids without any signs of dysphagia or aspiration. He was started on proton pump inhibitors to alleviate signs of reflux. His respiratory symptoms, cough and aspiration pneumonia improved by day three of admission and he was continued on oral antibiotics for a total of five days post-discharge. He was scheduled for outpatient follow-up to further investigate and manage suspected ineffective oesophageal motility or oesophageal aperistalsis.

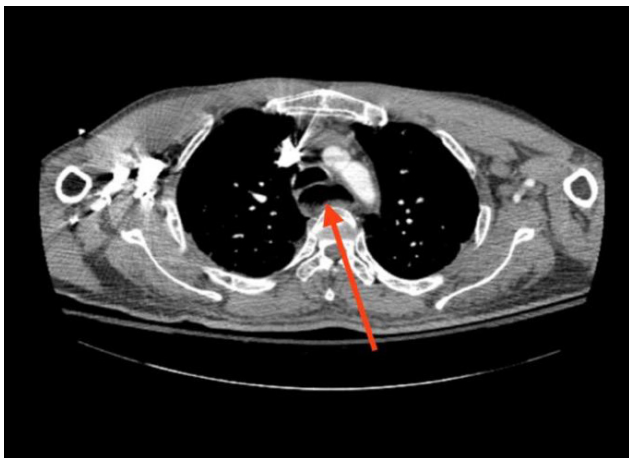

Figure 1. A computed tomography scan of the chest showing dilated oesophagus with fluid secretion (red arrow).

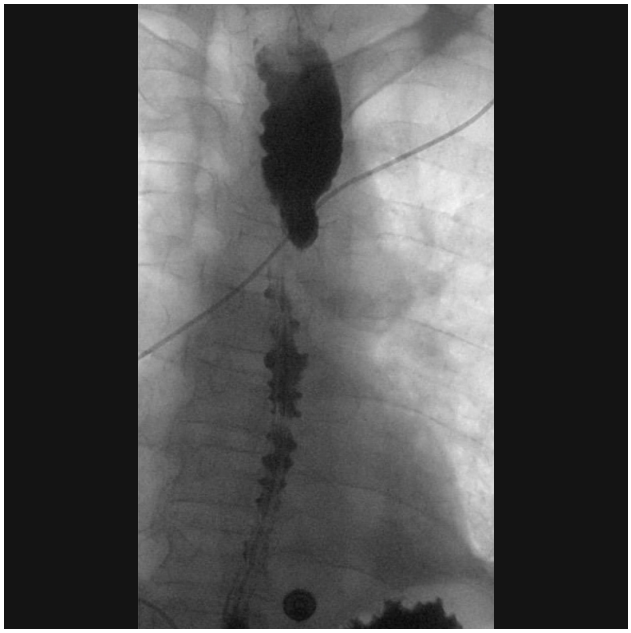

Figure 2. Barium oesophagram showing moderate dilatation in the upper oesophagus and severe oesophageal dysmotility; the dye did not clear through the oesophagus throughout the procedure.

DISCUSSION

CMT is the most common hereditary neuromuscular disorder[2] that typically affects peripheral nerves, leading to motor and sensory deficit[3]. While CMT's classic presentation includes limb weakness and sensory loss, evolving studies and case reports have shown that even though rare, patients may also experience autonomic dysfunction with respiratory and gastrointestinal complications, notably oesophageal dysfunction and dysphagia[4,5]. This review summarises current literature on the prevalence, pathophysiology and clinical implications of oesophageal involvement and dysphagia in CMT patients. Oesophageal motility is operated by a complex interplay of neural control and disturbances

in nervous supply, which can lead to various dysfunctions. Patients with CMT can experience autonomic dysfunction, which could extend to gastrointestinal motility issues leading to impaired coordination of peristalsis and resultant dysphagia; similarly, involvement of the diaphragm can lead to respiratory compromise[1,4].

Patients with gastrointestinal involvement often present with symptoms of dysphagia, which can manifest as difficulty swallowing solids or liquids[6], a sensation of food getting stuck or even regurgitation. One of the first reports of this uncommon association in 2009 highlights that dysphagia is a common but underrecognised issue in CMT, often attributed to oesophageal motility disorders[7]. Evaluation of oesophageal motility disorders in CMT patients typically involves a combination of clinical evaluation and diagnostic tests. In general, evaluation for dysphagia includes endoscopy, barium swallow and manometry. Endoscopic evaluation is useful to rule out structural abnormalities and assess complications such as oesophagitis[8,9]. Oesophageal manometry is the gold standard for assessing motility patterns, revealing abnormalities such as absent peristaltic amplitude in achalasia or disordered contraction pattern as in hypercontractile motility disorder[8]. However, these findings may not be the expected results in a patient with CMT. Instead, ineffective oesophageal motility would be anticipated or oesophageal aperistalsis, according to the Chicago Classification 4.0[7].

The clinical implications of oesophageal dysfunction in CMT patients are significant. Patients with CMT should be routinely screened for dysphagia and reflux disease as these can be indicators of oesophageal motility disorders, given the potential impact on nutrition and overall health. Furthermore, healthcare providers must be vigilant in monitoring and managing these complications to prevent severe outcomes of dysphagia such as aspiration pneumonia, malnutrition and an overall decline in quality of life[7]. Management of dysphagia in CMT patients requires a multidisciplinary approach and focuses on mitigating symptoms due to the limited therapeutic strategies available. These may include dietary modifications including the use of thickened liquids and softer food textures, swallowing therapy and pharmacological interventions to enhance motility such as prokinetic agents[6]. The use of proton pump inhibitors to manage gastroesophageal reflux may be employed to manage concomitant gastroesophageal reflux, which can exacerbate dysphagia symptoms[10].

CONCLUSION

Oesophageal dysfunction and resulting dysphagia are a critical yet often underrecognised complications in patients with CMT disease. The pathophysiological mechanisms underlying this dysfunction are complex, involving both motor and autonomic neuropathies. Clinicians must remain vigilant in identifying and managing dysphagia and related gastrointestinal symptoms in CMT patients to enhance their quality of life and prevent serious complications. Future research should focus on further elucidating the prevalence and specific mechanisms of oesophageal dysfunction in this population, as well as evaluating targeted interventions for management.

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