CASE REPORT

DYSPHAGIA AS AN UNCOMMON PRESENTATION OF MIXED CONNECTIVE TISSUE DISORDER: A CASE REPORT

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ABSTRACT

Mixed Connective Tissue Disorder is an autoimmune condition characterized by features of multiple connective tissue diseases. Dysphagia is a rare presentation of Mixed Connective Tissue Disorder and is not commonly reported. A unique and challenging case report to find out the cause of dysphagia as a Mixed Connective Tissue Disorder in a patient at a tertiary care Centre is presented. Early recognition, accurate diagnosis and appropriate management are crucial for optimal outcomes in such cases.

INTRODUCTION

Dysphagia may be a sensation that suggests abnormal movement of food bolus the oropharynx to the stomach, a lack of pharyngeal sensation or various other inadequacies of the swallowing mechanism. While dysphagia can be caused by many conditions, it is crucial to consider rare causes of dysphagia in the diagnostic process. MCTD is a rare disorder with an estimated incidence 2.1 per million per year. Almost any organ system can be involved, with esophageal symptoms being among the most common presentations (45–80%). Heartburn (48%) and dysphagia (38%) are the most common gastrointestinal symptoms reported in MCTD, but many patients may be asymptomatic.

Here we present a case of rare cause of dysphagia which highlights the importance of keeping rare cause in mind during diagnosis, as they can be challenging to manage and have significant Morbi-mortality rates. Furthermore, this case raises awareness of the ongoing discussion on the prognosis of this rare cause if dysphagia.

CASE REPORT

A 30-year-old female presented with difficulty swallowing solid food that was insidious in onset, gradually progressive, continuous, aggravated by solid food, and did not improve despite medication. She also complained of itching and rashes (Figure 1) on and off, irregular in shape, 1 cm in size, aggravated during the day and relieved at night. She also complained of mouth dryness, with no known aggravating or relieving factors. She denied any other medically relevant illness. There was no family history of esophageal disease. Her vitals parameters were consistent. The CNS examinations were normal.

Figure 1: Rashes on Arm
Figure 2: Upper Gastrointestinal endoscopy showing normal gastroesophageal junction

Figure 3: Nasal endoscopy showing normal mobile vocal cord with no pooling of saliva(abduction)

Figure 4: Nasal endoscopy showing normal mobile vocal cord with no pooling of saliva(adduction)

Obstructive causes were excluded after performing an upper gastrointestinal endoscopy (Figure 2), nasal endoscopy (Figure 3-4) and cervical computed tomography, while central neurogenic caused, mainly stroke and brainstem lesion, were excluded after a neurology consultation.

The patient was evaluated by rheumatology and found to have anti u1-Sn RNP/Sm RNP 14.0, Anti SSA/Ro 79.0, Anti Ro-52(SSA) 84.0, Complement 3(C3) 0.88gm/l (Table 1) which was diagnosed with MCTD based on her symptoms and laboratory findings. The patient was started on steroids and immunosuppressive drugs, which improved her symptoms of difficulty swallowing and skin rashes.

Table 1: Nuclear antigen antibody test

<table>
<thead>
<tr>
<th>S.N</th>
<th>Nuclear antigen antibodies</th>
<th>Result (CO/S) Ratio</th>
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<tbody>
<tr>
<td>1</td>
<td>Anti U1-snRNP/Sm RNP</td>
<td>14.0</td>
</tr>
<tr>
<td>2</td>
<td>Anti SSA/RO</td>
<td>79.0</td>
</tr>
<tr>
<td>3</td>
<td>Anti RO 52(SSA)</td>
<td>84.0</td>
</tr>
<tr>
<td>4</td>
<td>Complement 3</td>
<td>0.88</td>
</tr>
</tbody>
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DISCUSSION

MCTD is associated with esophageal dysmotility, GERD, and dysphagia. It can be subclinical at first, and up to one-third of patients with abnormal manometry tests are asymptomatic.4,5

The pathogenesis for dysmotility in patients with MCTD is unknown. One hypothesis is that autoantibodies attack smooth muscle tissue in the esophagus as well as ganglion cells in the Auerbach plexus, vascular walls, muscular tissue, and squamous epithelium contributing to the dysphagia.6 Postmortem analysis has shown histopathologic changes more severe in the lower two-thirds of the esophagus and in the inner circular muscular layer of the esophagus.7

Skin involvement occurs in most patients with Raynaud’s phenomenon as the presenting feature, but hypopigmentation/hyperpigmentation and scleroderma can also be present, similar to our patient’s presentation. Vasculopathy is similar to Systemic sclerosis and is characterized by bland intimal proliferation and medial hypertrophy that affects small and medium-sized vessels, also responsible for pulmonary arterial hypertension.6 Cardiovascular involvement varies between 11% and 85%, depending on the method used to detect.8 Pulmonary abnormalities are found in up to 85% of patients, such as interstitial lung disease and pulmonary arterial hypertension.6 Gastrointestinal disease is common (66%–74%) and often represents a major feature of overlap with Systemic sclerosis.9 In our case dysphagia was the primary main clinical symptoms and significantly impact the patient’s quality of life. Regarding diagnosis of MCTD, most physicians would agree that it should be considered in an anti-U1-RNP-positive patient presenting with Raynaud’s phenomenon, ‘puffy hands’ and at least two of the following clinical features: arthritis, myositis, leucopenia, esophageal dysmotility, pleuritis, pericarditis, interstitial lung disease or pulmonary arterial hypertension.9 Overall, diagnosing MCTD in clinical practice is an issue of pattern recognition and clinical decision.

CONCLUSION

In conclusion, we report a rare case of a patient newly diagnosed with MCTD after presenting with dysphagia symptoms. Laboratory, imaging, and clinical symptoms were used to determine the diagnosis of MCTD. A high suspicion for rheumatologic etiologies should be maintained when a patient presents with dysphagia and the appropriate clinical syndrome. It highlights the importance of a multidisciplinary in the diagnosis of dysphagia in rare conditions like MCTD.
REFERENCES:


