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Letters to the Editor

Utility of surgical myotomy in the dysphagia due to oculopharyngeal dystrophy

Key words: Oculopharyngeal muscular dystrophy. Cricopharyngeal myotomy. Dysphagia.

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Dear Editor,

Oculopharyngeal muscular dystrophy (OPMD) is a rare hereditary myopathy that affects mainly the levator palpebrae and the constrictor pharyngeal muscles, and causes severe dysphagia. It can be treated effectively by surgical cricopharyngeal myotomy, as demonstrated in the case presented below.

Case report

The case was a 69-year-old male suffering from dysphagia to solids and liquids, fever, respiratory secretions, weight loss, bilateral ptosis and dysphonia. Aspiration pneumonia was diagnosed by CT scan. After ruling out neurological focality, the electromyogram revealed a myopathic pattern with acute activity at rest.

The endoscopy failed to reach the esophagus. Videofluoroscopy found adequate tongue propulsion, an elevated epiglottis and an opened laryngeal vestibule (swallow aspiration), inactivity of constrictors causing abolition of the pharyngeal dynamics, absence of upper esophageal sphincter (UES) aperture before swallowing attempts and contrast retention in valleculae and pyriform sinuses (post-swallow aspiration). Manometry showed a normal resting UES tone, maximum pressure peaks of 230 mmHg and adequate pharyngosphincteric coordination, with an "ineffective motility" type disorder in the esophageal body.

Due to a significant deterioration of the patients' health and a genetic study that supported a diagnosis of OPMD, we decided to perform surgery after nutritional optimization with feeding gastrostomy. Extramucosal cricopharyngeal myotomy was performed (Fig. 1). Dysphagia was totally relieved and the patient was discharged on the seventh day, with oral intake, without tracheobronchial relapse symptoms after five months of follow-up.

Discussion

The OPMD raises a diagnostic and therapeutic challenge with respect to other more common causes of oropharyngeal dysphagia (1). Mutations in the PABPN1 gene (also called PABP2) on chromosome 14 (Cr14q11.2-q13) confirmed the diagnosis (2).

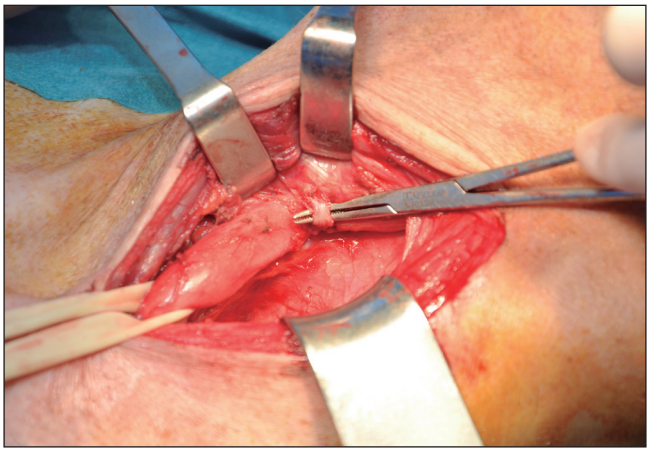

Fig. 1. We performed a left oblique cervicotomy. Then we dissected the esophagus (penrose drain in its caudal end). Myotomy starts by section of cricophayngeal muscle (marked with the dissecting forceps), and must be prolonged to the lower portion of the inferior pharyngeal constrictors and the upper centimeters of the cervical esophagus.

In the absence of curative treatment, the multidisciplinary approach aims to improve quality of life and reduce complications. Functional examinations, especially videofluoroscopy, allow the identification of the most appropriate therapeutic strategy for dysphagia (1): rehabilitation techniques (1), botulinum toxin injection (4), dilation (4), endoscopic or surgical UES myotomy. The latter has been shown to be effective in patients with OPMD (3-5).

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