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BRIEF COMMUNICATION

Cricopharyngeal Myotomy in the Treatment of Oculopharyngeal Muscular Dystrophy☆

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KEYWORDS
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Abstract Oculopharyngeal muscular dystrophy (OPMD) is an autosomal dominant myopathic disease which provokes oropharyngeal dysphagia, palpabral ptosis and proximal limb weakness. It is the abnormal expression of the GCG triplet in the *PABPN1* gene on chromosome 14 that causes this disease. The study of the oropharyngeal dysphagia that these patients suffer from should include upper gastrointestinal endoscopy, barium video-radiology and oesophageal manometry. Genetic study confirms the diagnosis. We report 6 patients (3 of whom were siblings) referred to our department with a confirmed diagnosis of OPMD, who underwent cricopharyngeal myotomy to achieve normal swallowing.

PALABRAS CLAVE
Distrofia muscular oculofaríngea;
Disfagia;
Tratamiento;
Miotomía del cricofaríngeo

Miotomía del cricofaríngeo en el tratamiento de la distrofia muscular oculofaríngea

Resumen La distrofia muscular oculofaríngea (DMOF) es una enfermedad hereditaria autosómica dominante que causa disfagia orofaríngea, ptosis palpebral y debilidad muscular proximal. Es causada por una expresión anormal del triplete GCG del gen *PABPN1*, situado en el cromosoma 14. El estudio de la disfagia orofaríngea que sufren estos pacientes se basa en la historia clínica, la endoscopia digestiva alta, la radiología con contraste baritado y la manometría esofágica. El diagnóstico definitivo se confirma con el estudio genético. Presentamos 6 casos, 3 de ellos de una misma familia, remitidos a nuestro departamento con el diagnóstico confirmado de DMOF, los cuales se sometieron a una miotomía del cricofaríngeo para conseguir una deglución normal.

Introduction

Oculopharyngeal muscular dystrophy (OPMD) is a late-onset autosomal dominant muscular illness. Progressive weakness of the ocular musculature was described by von Greafe in 1868 as ''external progressive ophthalmoplegia''.[1]

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Table 1 Clinical and Epidemiological Characteristics of the Patients.

Patient	Gender	Age	Symptoms	Personal History
1	Female	67	Dysphagia +++ Operated palpebral ptosis Proximal muscle weakness +	Of no interest
2	Female	71	Dysphagia +++ Operated palpebral ptosis Proximal muscle weakness +	Of no interest
3	Female	72	Dysphagia ++ Palpebral ptosis	Of no interest
4	Male	78	Dysphagia ++ Palpebral ptosis	Raised blood pressure
5	Male	58	Dysphagia ++ Palpebral ptosis	Of no interest
6	Male	57	Dysphagia ++ Palpebral ptosis	Of no interest

+: mild symptom; ++: moderate symptom; +++: severe symptom.

OPMD causes palpebral ptosis, oropharyngeal dysphagia and proximal weakness in the limbs. OPMD was finally described by Taylor in 1915.[2] The incidence and prevalence of this illness are very low, but cases have been reported all over the world. The population most affected by OPMD is to be found in the Quebec region of Canada, with a prevalence estimated at 1:1000[3] and in Israeli immigrants, with an estimated prevalence of 1:600.[4] OPMD is caused by the short expansion of the repeated triplet (GCG) 8–13 in the *PABPN1* gene located in chromosome 14.[5] In this paper, we report on 6 patients, 3 of them from the same family, referred to our department with a confirmed diagnosis of OPMD for the performance of cricopharyngeal myotomy (CM) in order to improve swallowing.

Materials and Methods

We conducted a prospective observational study of 6 patients diagnosed as having OPMD and referred to our department for assessment of surgery as treatment for their symptoms. The diagnosis had been genetically confirmed in all patients. Following surgery, they were monitored via the Otorhinolaryngology Out-Patients Clinic. Patients' clinical and epidemiological characteristics are listed in Table 1.

During the surgical procedure, a sample of the cricopharyngeal and sternocleidomastoid muscles was taken from each patient for study by pathologists. These samples were subjected to the normal tests for muscles (haematoxylin-eosin, Gomori's trichromic test, Oil Red O and PAS), as well as special histoenzymatic techniques (COX, SDH, NADH and ATPases 9.4, 4.6 and 4.3) (Figs. 1–3).

Results

CM was performed on all 6 patients. The approach consisted in a right cervicotomy, after placement of an inflated Foley catheter at the level of the Upper Oesophageal Sphincter (UOS), followed by exposure and identification of the cricopharyngeal muscle, and the performance of myotomy on the latter. Patients were discharged without complications the day after the procedure and were monitored through the ENT Out-Patient Clinic, with a minimum follow-up of 18 months and a maximum of 37 months. The improvement in dysphagia was moderate in 2 patients (33.3%) and considerable in the other four (66.6%). The results obtained and the review fibroendoscopic examinations are shown in Table 2.

The pathology study of the cricopharyngeal muscle revealed severe atrophy of the muscle fibres, with fibrosis and adipose substitution, with identification of vacuoles rimmed with basophilic material, characteristic of OPMD; there was neither necrosis nor inflammatory infiltrates. There were much more discreet in the sternocleidomastoid muscle, with no fibrosis but some atrophic fibres with rimmed vacuoles.

Discussion

OPMD is an autosomal dominant myopathic disorder causing palpebral ptosis, oculopharyngeal dysphagia and proximal muscle weakness. This disease is included within those caused by triplet repetition. OPMD is caused by the expansion of a GCG triplet located on the first exon of gene 1 in the polyadenylate binding protein (*PABPN1*), in chromosome 14 (14q11.2–q13).[5] Nonetheless, cases have been described with other mutations and other phenotypes, albeit rarely.[6] Cases of OPMD have been reported all over the world. The 2 largest populations of these individuals can be found in Canada, due to a family of French origin that emigrated to Canada in 1634,[3] and among Bukhara Jews who emigrated to Israel from Uzbekistan.[4] Smaller populations have been described in Europe and in the United States of America. Clinical signs include palpebral ptosis, dysphagia and proximal muscle weakness. Palpebral ptosis may precede or appear together with dysphagia in most patients; muscle weakness may appear after ptosis and dysphagia.

Swallowing is a neurophysiologically complex action. The correct operation of the UOS is an essential part of correct swallowing; its alteration may lead to potentially life-threatening dysphagia. Barium meals and video-endoscopy are 2 major tools for determining the severity of dysphagia.