CASE STUDY

Oculopharyngeal Muscular Dystrophy: A Case Report and Review of the Literature∗

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Abstract Oculopharyngeal muscular dystrophy is an infrequent, not widely known entity. Of genetic origin, it usually shows up in the 5th or 6th decade of life. Most cases are referred directly to the gastroenterologist by their general practitioner and not to the otolaryngologist, so it is essential to be aware of this disease to suspect it. We report a case diagnosed and treated in our hospital and we review the literature.

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Distrofia muscular óculo-faríngea: presentación de un caso y revisión de la literatura

Resumen La distrofia muscular óculo-faríngea es una entidad infrecuente y poco conocida, genéticamente determinada, que suele presentarse en la 5.ª o 6.ª década de la vida. Casi siempre estos pacientes son remitidos directamente al digestólogo desde su médico de familia y no con tanta frecuencia al otorrinolaringólogo, por lo que el conocimiento de esta enfermedad es fundamental para sospecharla. Presentamos un caso diagnosticado y tratado en nuestro hospital y revisamos la literatura al respecto.

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Case Report

A 78-year-old female attended our clinic complaining of dysphagia for solids—especially large pills—and dysphonia lasting for approximately two years. She did not report any choking incidents.

She was being treated with anti-hypertensives, hypcholesterolaemic agents and alendronate as therapy for the osteoporosis she was suffering from.

At the general level, her considerable thinness was noteworthy (42 kg and 1.69 m; BMI=14.71), but the patient reported that she had always had the same build and acknowledged a certain tendency to reject several foodstuffs for the last 30 years.

There was evidence of marked bilateral palpebral ptosis forcing the patient to hold her head backwards at a slight angle during the interview (Fig. 1).


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Figure 1 Marked palpebral ptosis. It is possible to appreciate a tendency to contract the brow muscle to raise the eyelids.

Clinical examination using a flexible nasofibrolaryngoscope revealed a large retention of secretions in both pyriform sinuses and retrocricoid area. Laryngeal mobility and the examination of cranial pairs were normal.

A blood test and biochemical analysis with nutritional profile were requested and these only detected mild ferrogenic anaemia, so oral treatment with iron was begun.

The collaboration of the Digestive Department was requested due to a suspicion of an oesophageal functional disorder.

Oesophageal-gastro-duodenal transit could not be completed due to the occurrence of bronchoaspiration resolved with medical treatment. Although aspiration occurred during the examination, the patient does not normally suffer from aspirations. The oesophageal gastroscopy detected a hernia in the hiatus and an extensive pylorus, as well as abundant secretions at the oesophageal outlet.

Oesophageal manometry was later performed and the conclusion reached was a pharyngeal motility disorder with severe hypotonia, hypertonia of the cardia with incomplete relaxations and alteration of the motility of the oesophageal body with 30% of simultaneous and others not transmitted (Fig. 2).

In view of the palpebral ptosis observed, an electromyogram (EMG) was performed on the limbs and was compatible with primary involvement of the muscle fibres, supporting the diagnosis of oculopharyngeal muscular dystrophy (OPMD).

The gene study requested identified a pathological expansion of 9 GCG triplets in one of the alleles of the PABPN1 gene, and 6 normal-sized repetitions in the contrary allele. This result is compatible with the diagnosis of OPMD. The technique used was PCR-based amplification of the expansion area.

Six months after starting a diet with thickeners, a weight gain of 6 kg was achieved. Residues remaining after the post-swallowing phase are what cause the patient to require pharyngeal clearing. Foods with the consistency of a homogeneous puree or creme caramel, heavier and with a tendency to slide more easily, provide greater comfort when eating with this kind of pathology.

In addition, alendronate was withdrawn to reduce the risk of erosive lesions on the mucosa of the oesophagus because of its corrosiveness.

The patient is currently carrying out a high level of physical activity for her age without experiencing asthenia. She has refused any other invasive therapeutic measure so no other options will be proposed so long as she maintains a normal nutritional status and does not present aspirations.

Discussion

OPMD is an autosomal dominant genetic disease in which an expansion of the GCG triplet has been identified on exon 1 of the PABPN1 gene. A greater tendency is seen in families with a French-Canadian background. It involves an oropharyngeal motor dysphagia in which there is hypomotility of the pharynx, an incomplete relaxation of the sphincter during pharyngeal contraction and/or pharyngeal-sphincteral incoordination. It is frequently accompanied by regurgitation of the bolus towards the nasopharynx and respiratory symptoms due to laryngeal aspiration. In the final stages of the illness, weakness may spread to the muscles in the limbs or the rest of the face and may even cause double vision or weakness in the voice.1

<table>
<thead>
<tr>
<th>LOS-LOS summary</th>
<th>LOS-LOS relaxation summary table</th>
<th>LOS-LOS pressure at rest summary table</th>
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<td>Lower edge of the LOS (cm)</td>
<td>Residual pressure (mm Hg)</td>
<td>Residual pressure (mm Hg)</td>
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<td>43.8</td>
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<tr>
<td>Upper edge of the LOS (cm)</td>
<td>Relaxation index (%)</td>
<td>Pressure at rest (mm Hg)</td>
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<td>6.5</td>
<td>-</td>
<td>-</td>
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<tr>
<td>Abdominal length (cm)</td>
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Figure 2 Oesophageal manometry showing severe hypotonia, hypertonia of the cardia with incomplete relaxations and alteration in the motility of the oesophageal body with 30% simultaneous waves and others not transmitted.
The main causes of oropharyngeal motor dysphagia are: neuromuscular pathology, diseases of the central nervous system (Parkinson's disease, multiple sclerosis, vascular accidents), peripheral neuropathies in cranial nerves IX, X and XII, illnesses of the neuromuscular junction (myasthenia gravis) and muscular diseases (polymyositis and muscular dystrophies). A functional study of swallowing is fundamental in order to reach a definitive diagnosis. Thus, the findings of the oesophageal manometry in OPMD include: hypertonia of the upper oesophageal sphincter (UOS), not in all cases inco-ordination of the UOS and pharynx, weak pharyngeal pressure and severe pharyngeal hypotonia. In our case, no hypertonia of the UOS was detected, with the hypotonic component predominating instead.

Although videofluoroscopy is not available at our centre, in the pharyngeal phase of this test we would observe a homogeneous, symmetrical post-swallowing residue in both pyriform sinuses as a sign of weak contraction of the pharyngeal constrictors, thus diminishing pharyngeal clearance. This sign is very frequent in neuromuscular illnesses and predisposes for post-swallowing aspirations.

Pharyngo-oesophageal manometry and videofluoroscopy may also be useful for indicating a direct procedure on the UOS as the probabilities of success are greater if hypertonia or a lack of relaxation is observed.

Treatment for OPMD was gradually introduced in view of the symptoms, which also ensue insidiously. The absence of effective treatments means that its is handled similar to other degenerative neurological diseases, facilitating feeding through postural measures or changes in food texture. Medication that might increase dysphagia must be avoided, whether through a directly caustic effect or other mechanisms. In cases of hypertonia of the UOS, nifedipine (20–40 mg sublingual) or isosorbide dinitrate (5 mg) can be used before meals. Other feasible interventionist procedures in case of hypertonia of the UOS are surgical or endoscopic myotomy or the injection of botulinum toxin. The indication of percutaneous gastrostomy is reserved for cases in which the measures mentioned fail or those in which the risk of aspiration is very high.

The thickeners used may be gelled water or syrups. It is necessary to avoid eating foodstuffs with heterogeneous consistencies as they make choking and aspiration easier.

Conclusions

OPMD may easily go unnoticed unless it is known about and suspected. Rehabilitation and dietary adaptation are fundamental links in the therapeutic chain. Malnutrition and/or aspiration are potential and harmful complications that may reduce the life expectancy of these patients. Gene repair therapy or cell therapy are opportunities to achieve a cure for this condition.

Conflict of Interest

The authors have no conflict of interests to declare.

References