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LETTER TO THE EDITOR

Myogenic abnormalities in intensive care can hide an uncommon diagnosis

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Introduction

Diagnosis of muscle disorders is often challenging and requires a meticulous comprehensive work-up. We describe a case of rapidly progressive oculopharyngeal muscular dystrophy (OPMD) misdiagnosed as critical illness myopathy (CIM) due to the aspecific nature of its main symptoms. Through this report we aim to highlight the importance of careful medical history and meticulous physical examination to avoid neglecting infrequent and unfamiliar muscle diseases.

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Case report

A 60-year-old woman was admitted to intensive care unit because of acute aspiration pneumonia complicated of acute respiratory failure, requiring intubation and mechanical ventilation. Despite favourable response to antibiotics, she was difficult to wean from mechanical ventilation, resulting in 3 weeks hospitalization in intensive care. Since she has reported daily alcohol and nicotine abuse over years, the possibility of malignancy process was considered but cervical and chest computed tomography, oesophageal gastroscopy and laryngoscopy ruled it out. However, laryngoscopy showed incomplete glottic closure and impaired vocal cord mobility.

An electromyography (EMG) was performed to help the diagnostic process: short-duration polyphasic motor unit potentials were found bilaterally in *brachialis biceps*, *anterior tibialis* and *extensor hallucis longus* muscles; conduction nerves velocities and potential amplitudes (sensory and motor nerves) were normal. CIM was then diagnosed because of proximal limbs weakness in a context of predisposing conditions (namely sepsis and failure to wean from the ventilator) and according to the myogenic abnormalities found during the EMG.

She was referred to our department 3 months later for follow-up. The general medical history was unremarkable, except for scoliosis. Careful history revealed progressive dysphagia since 1 year causing weight loss of 10 kg (BMI 15 kg/m^2). She was also complaining of dyspnea over months and pulmonary function testing showed reduced inspiratory muscle strength with a maximum inspiratory pressure (PImax) of 13 cmH_2O (predicted normal value 79.5 cmH_2O) and a sniff nasal inspiratory pressure (SNIP) of 30 cmH_2O (predicted normal value 83.5 cmH_2O). Physical examination showed bilateral ptosis which had

never been noticed by her family or practitioners before neurological assessment, bilateral ophtalmoparesis, nasal speech and facial muscles weakness. Reflexes were not decreased. Blood tests were normal, especially creatine kinase concentration.

Family history revealed that her mother had died of aspiration pneumonia and we surprisingly observed that her daughter (32-year-old) presented ptosis and nasal speech, which led us to consider hereditary muscular dystrophy, especially as EMG did not show any denervation despite myogenic abnormalities. Molecular genetic analysis identified a pathological expansion of 12 GCG triplets in one of the alleles of the *PABPN1* gene, confirming oculopharyngeal muscular dystrophy OPMD. She agreed to percutaneous endoscopic gastrostomy placement to improve nutritional situation.

Discussion

OPMD is a rare inherited autosomal dominant adult-onset disease due to expansion of GCG triplets in *PABPN1* gene on chromosome 14q11 with an estimated prevalence of 1:100,000 in Europe [1]. Onset typically occurs in the fifth to sixth decade and clinical diagnosis relays on the combination of ptosis, as the first and cardinal symptom, usually followed by dysphagia and proximal limb weakness [1]. Disease course is always progressive but various clinical presentations can be observed. Swallowing difficulties can be delayed and arise up to 17 years after ptosis [2] but have rarely been reported as the revealing manifestation of OPMD [1, 2]. Extraocular and facial muscles can also be later gradually involved [3].

Although pulmonary involvement is not considered as a feature of OPMD, our patient complained of respiratory symptoms consistent with results of pulmonary function testing. Reduction of forced expiratory volume >20% has been reported in a series of thirteen patients with OPMD who did not complain of respiratory symptoms [1].

EMG classically demonstrates signs of myopathic process [4] and although it is not mandatory for diagnosis (due to absence of typical features), it can be helpful in differential diagnosis which consists in myasthenia gravis, myotonic dystrophy and amyotrophic lateral sclerosis. Here, the alert signs for the differential diagnosis of CIM were the absence of spontaneous fibrillation potentials, atypical for acute CIM, and the absence of neuropathy frequently combined to CIM.

Due to its insidious evolution and lack of familiarity, this myopathy is commonly under recognized. Misdiagnosis is frequent with many hospitalizations before accurate diagnosis as patients are first referred to otolaryngologists, gastroenterologists, ophthalmologists or intensivists. Practitioners should be aware of this combination of evocative symptoms and suspect the possibility of OPMD in patients with dysphagia and/or ptosis of unknown origin. Contribution of meticulous history and family history is essential to achieve appropriate diagnosis.

Despite the absence of current curative treatment, early diagnosis allows (1) appropriate management by preventing potentially life threatening complications of swallowing difficulties, providing dietary (or texture) adaptation and (2) genetic counselling. Improvement of dysphagia with cricopharyngeal myotomy has been reported to be effective but high recurrence is observed [5]; surgical correction for ptosis may also be provided.

Compliance with ethical standards

Conflict of interest The authors declare no conflict of interest related to this publication.

Ethical statement This article does not contain any studies with human participants or animals performed by any of the authors.

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References

1. Witting N, Mensah A, Køber L, Bungaard H, Petri H, Duno M, Milea D, Vissing J (2014) Ocular, bulbar, limb and cardiopulmonary involvement in oculopharyngeal muscular dystrophy. Acta Neurol Scand 130:125–130
2. Tondo M, Gámez J, Gutiérrez-Rivas E, Medel-Jiménez R, Martorell L (2012) Genotype and phenotype study of 34 Spanish patients diagnosed with oculopharyngeal muscular dystrophy. J Neurol 259:1546–1552
3. Werling S, Schrank B, Eckardt AJ, Hauburger A, Deschauer M, Müller M (2015) Oculopharyngeal muscular dystrophy as a rare cause of dysphagia. Ann Gastroenterol 28:291–293
4. Bouchard JP, Brais B, Brunet D, Gould PV, Rouleau GA (1997) Recent studies on oculopharyngeal muscular dystrophy in Quebec. Neuromuscul Disord 7:S22–S29
5. Coiffier L, Périé S, Laforêt P, Eymard B, St Guily JL (2006) Long-term results of cricopharyngeal myotomy in oculopharyngeal muscular dystrophy. Otolaryngol Head Neck Surg 135:218–222