Case Report

URF PUBLISHERS
connect with research world

World Journal of Surgery and Surgical Case Reports

https://urfpublishers.com/journal/surgery-and-surgical-case-reports

Vol: 1 & Iss: 2

The DAM Effect: A Case Report on Dysphagia in a Patient with Oculo-Pharyngeal Muscular Dystrophy

Alice Di Battista[1], Federica Perpiglia[2], Marco Ruggiero[3]* and Alessandro Conforti[4]

[1]ASL Roma 3, Rehabilitation Unit, Centro Paraplegici Ostia, Rome, Italy

[2]AUSL Romagna, Rehabilitation Unit, Ravenna, Italy

[3]ASL Roma 2, Rome, Italy

[4]ASL Roma 4, Local Health Unit Roma 4, Civitavecchia, Rome, Italy

Citation: Di Battista A, Perpiglia F, Ruggiero M, Conforti A. The DAM Effect: A Case Report on Dysphagia in a Patient with Oculo-Pharyngeal Muscular Dystrophy. *World J Surg Surgical Case Rep*, 2025;1(2):51-53.

Received: 12 July, 2025; **Accepted:** 21 July, 2025; **Published:** 23 July, 2025

***Corresponding author:** Marco Ruggiero, ASL Roma 2, Rome, Italy, E-mail: marco.ruggiero@aslroma2.it

Copyright: © 2025 Ruggiero M, et al., This is an open-access article published in World J Surg Surgical Case Rep and distributed under the terms of the Creative Commons Attribution License, which permits unrestricted use, distribution, and reproduction in any medium, provided the original author and source are credited.

ABSTRACT

Oculo-Pharyngeal Muscular Dystrophy (OPMD) is a rare form of muscular dystrophy typically occurring in individuals over 50. Its primary symptoms include eyelid ptosis, dysphagia and proximal limb weakness. This case report describes the clinical presentation, challenges and rehabilitation approach for a 61year-old female patient with OPMD. The aim is to highlight an atypical manifestation of dysphagia, referred by the patient as a "dam effect," and to emphasize the importance of a multidisciplinary rehabilitation strategy. Despite the lack of specific guidelines in the literature, this case highlights the fundamental role of personalized treatment in enhancing the patient's quality of life.

Keywords: Oculo-pharyngeal muscular dystrophy, Dysphagia, Proximal limb, Rehabilitation

1. Introduction

Oculo-Pharyngeal Muscular Dystrophy (OPMD) is an intractable inherited myopathy caused by a genetic anomaly. It is considered a rare disease and its incidence varies among different ethnic groups, with an incidence ranging from 1:100,000 to 1:1,000,000 in Europe[1,2]. It has a late onset around the fourth-fifth decade of life. According to a recent review[3], the main symptoms of OPMD are ptosis, dysphagia and proximal limb weakness. Patients with OPMD may also experience extra-muscular symptoms such as deterioration of respiratory function, dementia, executive dysfunction and generalized fatigue.

The typical swallowing disorder is solid food becoming lodged in the throat[4]. Primary functional impairments with swallowing inefficiency (e.g., pharyngeal residue) and food aspiration have been identified[5,6], which can lead to life-threatening complications such as choking, aspiration pneumonia or malnutrition. Recent reports suggest that swallowing problems in OPMD are not limited to pharyngeal weakness, but that tongue strength and oral bolus control may also be reduced. Speech changes have also been documented, ranging from palatal weakness causing a nasal voice to articulation problems and reduced speech rate[2,6].

This clinical case was previously presented as a short communication at the 2022 ESLA European Speech and Language Therapy Association on May 28, 2022.

2. Case Presentation

A 61-year-old female high school teacher, diagnosed with OPMD in 2016, was referred to a Speech and Language Pathology (SLP) center for worsening dysphagia and asthenophonia. The patient's medical history included blepharoplasty in 2018 and ongoing management by a multidisciplinary team comprising a neurologist, physiatrist, physiotherapist and dietitian. Despite being underweight, the patient refused Percutaneous Endoscopic Gastrostomy (PEG) and relied on oral intake supplemented by high-calorie diets prescribed by a dietitian.

The patient reported a lump in her throat when eating solid foods, requiring subsequent boluses to advance the previous bolus into the esophagus.Consequently, the patient reported difficulty in managing the final bolus, which she often expelled by leaning her chest forward and using her hands. The "dam effect," as she literally described it, refers to the attempt to clear the last bolus with water, resulting in oropharyngeal filling with fluids that cannot pass through the esophagus and thus pose a significant problem.

The patient managed her diet by alternating between soft and bite-sized foods (IDDSI Level 6) and pureed foods (IDDSI Level 4) and sometimes minced and moist foods (IDDSI Level 5) depending on her fatigue level. Soft drinks (IDDSI Level 0) were consumed separately to avoid choking episodes.

2.1. Clinical findings

2.1.1 Clinical Swallow Evaluation

- Oral inspection revealed complete dentition, cleansed mucosa and minimal thick secretions in the oropharynx.
- Oral motor evaluation showed adequate strength and precision, except for velar motility deficits, causing nasal regurgitation.
- Reflex evaluation demonstrated a delayed pharyngeal swallow reflex but preserved cough reflex.
- Swallowing tests demonstrated:
  - Thin Drinks (IDDSI 0): Functional swallowing but preferred gelled water (IDDSI Level 4) during work for easier management;
  - Pureed Foods (IDDSI 4): Functional swallowing;
  - Soft and bite-sized foods (IDDSI 6): Slight delay in swallowing reflex initiation;
  - Poor laryngeal elevation in no pharyngeal stagnation sensation;
  - Regular Foods (IDDSI 7): Delayed swallowing (>10 sec) and sensation like vallecular stagnation.

2.1.2 Instrumental Evaluation

Fiberoptic Endoscopic Evaluation of Swallowing (FEES) revealed nasopharyngeal secretions, food residues in the vallecula and pyriform sinuses and compensatory head postures (chin tuck posture) required for bolus clearance (Table 1).

All the dysphagia tests administered confirmed that the severity of dysphagia was moderate. Swallowing-related quality of life was also assessed using the MDADI test, which revealed a physical rather than an emotional and functional impact. The patient is aware of the difficulty but seems able to manage it.

The SLP evaluation also included dysarthria assessment. The patient presents a nasal voice, articulation difficulties, particularly with velar sounds and a reduction in speech rate. This condition is aggravated by fatigue, leading to asthenophobia and pneumonic incoordination. The shortened Robertson test questionnaire was administered, which confirmed the qualitative findings. In Table 2, voice and articulation tests are reported.

Table 1: Dysphagia assessment.

Dysphagia Tests	Score
MASA Mann,2022	176 Mild Dysphagia
DOSS O'Neil, 1999	4 Mild moderate Dysphagia
MDADI Shindler, 2008	21 Dysphagia perceived as medium disability Global 2 Emotional 3 Functionals 5 Physical 11
P-SCORE Farneti, 2008	8 Moderate Dysphagia
PAS Rosenbek, 1996	2 Penetration food remains above the vocal folds not ejected

Table 2: Dysarthria assessment.

Voice And Dysarthria Tests	Score
GIRBAS	G1 I2 R0 B0 A2 S0
VHI Jacobson, 1997	P17 F11 E17
Robertson Dysarthria Profile mod. and adapted Italian version, 2015	Intelligibility 6/8 Breathing 8/12 Phonation 2/4 Diadocokinesis 18/24 Orofacial Musculature 60/64 Prosody 15/16 Articulation 10/12

2.2. Rehabilitation approach

2.2.1. Swallowing training:

- Compensatory posture (chin tuck posture) to improve bolus clearance.
- Muscular strengthening exercises (e.g., Masako Maneuver) for tongue, lips and velar muscles.
- High-intensity task-oriented swallowing practice.

2.2.2. Dysarthria management:

- Pneumo-phonic coordination exercises to reduce asthenophonia.
- Articulation and velar sound exercises.
- Vocal hygiene education.
- Counselling to help the patient obtain an exemption from work by reducing vocal effort.

The patient demonstrated improved swallowing, reduced nasal regurgitation and improved bolus management. She successfully adopted compensatory strategies, which improved her feeding confidence and quality of life. Dysarthria symptoms, including nasal voice and fatigue, were managed with pneumophonic coordination and articulation exercises.

3. Discussion

This case highlights the challenges in managing OPMD-related dysphagia, particularly in the presence of atypical manifestations such as those reported by the patient as a "dam effect." The findings highlight the importance of a multidisciplinary approach, involving neurologists, physiotherapists, dietitians and SLPs, to address the different needs of patients with OPMD[7,8].

Although the literature supports the role of rehabilitation in managing swallowing and articulation deficits[9,10], there is limited evidence describing specific treatment protocols for OPMD.

When evaluating this patient's therapeutic response, the role of fatigue should be considered as a factor that has a cumulative effect on the symptoms of dysphagia and dysarthria. The patient presented with asthenophonia and pneumophonic incoordination aggravated by fatigue, meaning that the treatment schedule and intensity must be adjusted based on energy status. Energy-saving measures, including scheduling therapy during peak alertness hours and including rest periods make treatment more effective and avoid the frustration resulting from worsening symptoms.

Another issue that emerged was the psychosocial burden of patients with OPMD. Despite her clinical difficulties, the patient continued to work as a teacher, a job that requires a lot of verbal communication. Her resistance to PEG and her subsequent dependence on oral feeding indicates a strong need for independence. While these decisions are laudable, they also demonstrate the need for psychological support during rehabilitation. Emotional resilience and counselling regarding disease progression, social identity and professional roles can improve overall well-being and promote adherence to treatment recommendations.

Finally, this case raises further questions about the need for unified yet adaptable intervention models for rare neuromuscular diseases. While general principles of dysphagia management can be applied, OPMD presents specific combinations of motor deficits that require assessment tools and personalized treatment pathways. The so-called dam effect, described by the patient, conveys a clinically essential yet underrecognized symptom, which could be used in the future to establish diagnostic or therapeutic guidelines. By recording these idiosyncratic presentations and combining them with functional findings, clinicians can contribute to the growing body of experiential knowledge base, leading to the definition of evidence-based best practices in the management of OPMD-related dysphagia.

4. Conclusion

This case report suggests how a multidisciplinary rehabilitation strategy can significantly improve the quality of life of patients with OPMD. Despite the rarity of the condition, detailed case reports like this may provide valuable insights into personalized interventions for dysphagia and dysarthria. Therefore, further studies are needed to establish evidence-based protocols for the management of OPMD.

5. References

1. Ruggiero M, Conforti A, Culcasi A, Mazzanti C, Sibahi G, Rani N, Sartini S. A focus on melorheostosis disease: a literature review and case report of femoral-acetabular impingement due to melorheostosis treated with surgical hip osteoplasty. Reumatismo. 2024;76(1).
2. Yamashita S. Recent progress in oculopharyngeal muscular dystrophy. J Clin Med. 2021;10(7): 1375.
3. Meola G, Sansone V, Rotondo G, et al. Oculopharyngeal muscular dystrophy in Italy. Neuromuscul Disord. 1997;7(1): 53-56.
4. Kroon RHMJM, Horlings CGC, de Swart BJM, et al. Swallowing, chewing and speaking: frequently impaired in oculopharyngeal muscular dystrophy. J Neuromuscul Dis. 2020;7(4): 483-494.
5. Agarwal PK, Mansfield DC, Mechan D, et al. Delayed diagnosis of oculopharyngeal muscular dystrophy in Scotland. Br J Ophthalmol. 2012;96(2): 281-283.
6. Knuijt S, Cup EH, Pieterse AJ, et al. Speech pathology interventions in patients with neuromuscular diseases: a systematic review. Folia Phoniatr Logop. 2011;63(1): 15-20.
7. Menendez Sepulveda JA, Izquierdo N. Oculopharyngeal muscular dystrophy: A case report from Puerto Rico. Cureus. 2024;16(7): 65766.
8. Infante JM, Nepomuceno BL. Choked: a case report of oculopharyngeal muscular dystrophy mimicking hypothyroidism from the Philippines. Cureus. 2023;15(6): 41025.
9. Tabor LC, Plowman EK, Romero-Clark C, et al. Oropharyngeal dysphagia profiles in individuals with oculopharyngeal muscular dystrophy. Neurogastroenterol Motil. 2018;30(4): 13251.
10. Palmer PM, Neel AT, Sprouls G, et al. Swallow characteristics in patients with oculopharyngeal muscular dystrophy. J Speech Lang Hear Res. 2010;53(6): 1567-1578.