Oculopharyngeal Muscular Dystrophy: a Case Report

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ABSTRACT

Objectives: To report rehabilitation management of oculopharyngeal muscular dystrophy (OPMD).
Study design: Case report.
Setting: Department of Rehabilitation Medicine, Maharat Nakhon Ratchasima Hospital.
Subject: A 72-year-old male who presented with progressive bilateral ptosis, hoarseness and swallowing difficulty, dysphagia.
Methods: Patient's medical records from 1st January 2018 to 31th December 2019 were reviewed and swallowing dysfunction was assessed by physical examination, a modified water swallow test and videofluoroscopy.
Results: Abnormal swallowing was found in the pharyngeal stage with decrease in laryngeal elevation and pharyngeal constriction. Penetration was seen after drinking a cup of water. OPMD was clinically diagnosed. Genetic confirmation of mutation in the PABPN1 gene was made. Nutritional assessment was performed. Swallowing rehabilitation program consisted of Shaker exercise and Mendelsohn maneuver to increase hyolaryngeal excursion, chin tuck for compensatory strategy and modified diet. The patient was discharged without complication. The swallowing program was continued and he was able to swallow safely.
Conclusion: OPMD is a rare disease causing dysphagia. Extensive family history-taking and physical examination leaded to diagnosis. Currently, there is no definite treatment for OPMD but proper swallowing rehabilitation management could improve swallowing dysfunction and improve patient's quality of life.

Keywords: oculopharyngeal muscular dystrophy, dysphagia, swallowing dysfunction, rehabilitation

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Introduction

Oculopharyngeal muscular dystrophy (OPMD) is one of nine types of muscular dystrophies.(1) Physical manifestation in patients begins with ptosis, which gets worse as time goes on. Later, the patients will have difficulty swallowing and at last will experience proximal limb weakness.(2) As the disease has its first manifestation in the voluntary muscles, OPMD is considered a rare disease. It is a genetic disorder, mostly from hereditary autosomal dominant which usually manifests in the fifth or sixth decade of life. However, some can also be an autosomal recessive disorder that happens rarely. It is difficult to diagnose as the manifestations happen at an advanced age and thus could be confused with aging conditions.(3) Therefore, the diagnosis requires genetic testing. Presently, the standard method for diagnosis is genetic study poly (A) binding nuclear protein 1 (PABPN1) gene in OPMD patients with autosomal dominant. It is found that there will be 12-17 repeats of expanded alleles. As for the autosomal recessive type, there are 11 repeats.(4)

The prevalence of autosomal dominant OPMD is high in Quebec province of Canada at the ratio of 1:1000 individuals. However, it was Isarael's Burkhara Jewish population that had the highest gene frequency at 1:600.(5) In Asian countries, there were some reports on the prevalence of OPMD, such as in China, Taiwan, Hong Kong, and Japan with reports on genetic confirmation in Southeast Asian countries such as Malaysia and Thailand.(6) Because it is a rare disease and not well-known outside Canada, specialists e.g. neurologists, otolaryngologists, have a potential to underdiagnose OPMD.(7) Though there has not been any exact report concerning the survival of untreated OPMD,(8) malnutrition and recurrent aspirations are important complications which can affect life expectancy of this group of patients if swallowing difficulty assessment is not included.(9) It is clear there will be great challenges in the diagnosis and treatment. Therefore, we would like to present a case with dysphagia in this rare genetic disease and results of swallowing assessment and rehabilitation program to improve swallowing function.

Case report

A 72-year-old Thai male with underlying type II diabetes and hypertension was presented to the hospital with hoarseness symptom for 6 years. Fiberoptic laryngoscope (FOL) examination was performed by an otolaryngologist, and presbylaryngis was diagnosed. He then lost to follow-up.

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One year later, he came with dyspepsia and bilateral ptosis. Gastritis was treated with proton pump inhibitors by an internist. His symptoms did not improve after being treated for two years. He was referred to gastro-enterologist for esophago-gastro-duodenoscopy (EGD) but findings were normal. Then the second FOL was done and showed only abnormal pooling of saliva at posterior cricoids, atrophic true vocal cord with good movement, and no mass in the oropharynx, pharynx and hypolarynx. The barium swallow showed normal visualized esophagus but large amount of barium suspension went into the trachea, thus the study was terminated. The patient needed oral feeding and his swallowing difficulty became worse. Finally, he was referred to a neurologist due to hoarseness, ptosis in both eyes and swallowing problems. He was asked to give a thorough record of his history as well as that of his extended family as shown in Figure 1.

His mother had an onset of symptoms, progressive dysphagia and bilateral ptosis, at the age of 57 years old, followed by an upper airway problem with stridor, and died of stroke at the age of 62 years old. His eldest brother had an onset of bilateral ptosis and ophthalmoparesis at the age of 66 years old and died of aspiration pneumonia at the age of 73. Another elder brother had uncertain onset of progressive bilateral ptosis and abnormal voice. The sixth brother had only bilateral ptosis and mild facial weakness at the age of 55 and died of metastatic brain cancer 7 years ago. The other two younger brothers presenting with bilateral ptosis, dysarthria, dysphonia and mild dysphagia at the age of 65 years old, were diagnosed with OPMD.

According to the above-mentioned family history, muscular dystrophy was suspected as the cause of his illness. The gene study was requested when he was referred to a rehabilitation physician (physiatrist) for assessment of swallowing difficulty. A DNA test reported heterozygous (GCG)9 or (GCN)13 expansion in the PABPN1 gene, resulting in an expansion of polyalanine tract from 10 to 13 residues in one allele, thus verifying the patient as having OPMD. Laboratory tests revealed normal serum albumin, creatinine and BUN levels, normal CBC counts and RBC indices, and HbA1C was 8.7%. His blood pressure was controlled. Muscle biopsy and serum creatine kinase level were not done.

Before admission for medical rehabilitation, his body weight decreased by 10% (from 55 kg to 48 kg) in the past year.

Dysphagia and ptosis progressed significantly. He reported difficulty swallowing of liquids primarily and then of solid foods but he denied ever experiencing choking events or lung infections. Physical examination showed good tongue strength and movement with no fasciculation or atrophy. Gag reflex was absent bilaterally. Soft palate elevation decreased on the left side and laryngeal elevation was more than one finger breadth when the patient performed a dry swallowing. Hoarseness was noticed but the patient could still communicate. The motor power was graded 5 in all extremities so he was independent in all daily activities and could walk without gait aid.

A modified barium swallow test showed decrease in laryngeal elevation and pharyngeal constriction and some residues after swallowing a tested food in an upright left lateral view (Figure 2), suggesting physiologic abnormalities in the pharyngeal stage, impaired relaxation of upper eso-phageal sphincter and inadequate airway protection. After doing a chin tuck, there was a decrease in residue after swallowing.

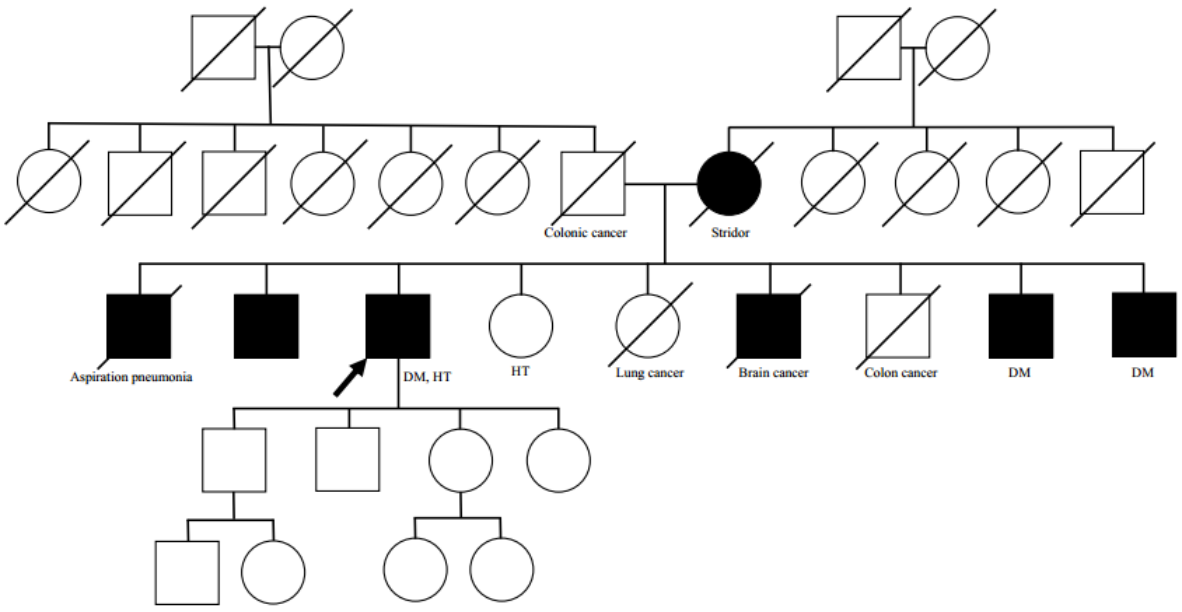

Figure 1. Pedigree of the patient's family: his mother (black circle) and six out of nine siblings including the patient (black square with black arrow) with diagnosis of oculopharyngeal muscular dystrophy (OPMD) indicating an autosomal-dominant transmission. The affection status of the maternal grandmother is unclear due to unrecognized history from the patient. The affection status of two generations after the patient are negative because the onset of symptom has not shown.

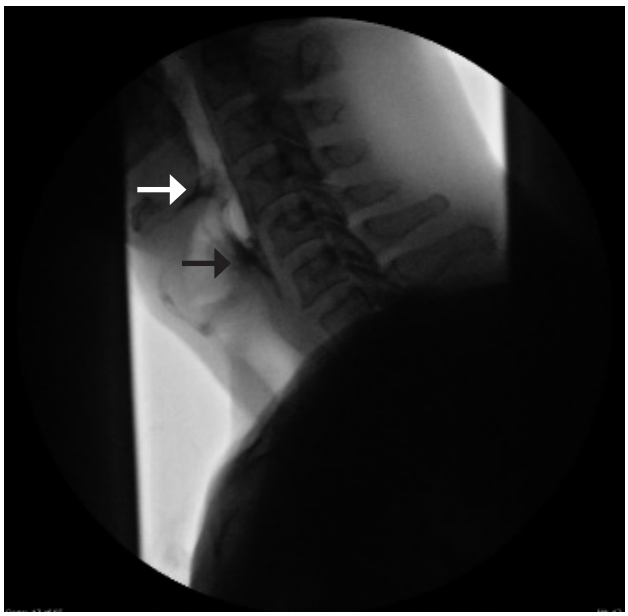

Figure 2. The modified barium swallow test shows residue in vallecular (white arrow) and pyriform sinus (black arrow).

Penetration was seen after drinking a cup of water and there was some residue after swallowing various food types such as liquid, paste, and solid. Oxygen saturation level did not change throughout the test.

During rehabilitation admission, the patient was trained to swallow using Mendelsohn maneuver and chin tuck position. In addition, Shaker exercise was advised to improve laryngeal elevation. Then he was discharged and asked to continue such program at home with a regular diet. After 2 months of home program, swallowing improved. Based on the Functional Oral Intake Scale (FOIS), swallowing was graded as level V, total oral diet with multiple consistencies, but required special preparations or compensations.

Discussion

Diagnosis of OPMD is difficult in Asian countries where the prevalence is low and even more difficult in cases of mild symptoms such as only ptosis or voice hoarseness. Multiple diagnoses and follow up visits were often made before getting a definite diagnosis of OPMD. In this case, he was firstly diagnosed by otolaryngologist because his main complaint was hoarseness. Only later when ptosis and swallowing difficulty increased, he was referred to a neurologist as a genetic neuromuscular disorder was suspected. Without thinking of this group of diseases, a misdiagnosis could lead to delayed treatments.

At present, treatments for OPMD are symptomatic, like those of degenerative neurological disease.[9] From previous researches,[1,3,9] swallowing difficulty started with solid food followed by liquid. This patient provided a different history as the problem started with liquid food and over 10% weight loss which led to a suspicion of pharyngeal muscle dysfunction. The modified barium swallow test showed impaired swallowing of both solid and liquid at the pharyngeal stage, similar to previous reports.[1,2,9,10,11] It was reported that 77.3% of patients with OPMD had food residues in vallecular and 90.1% had food residues in pyriform sinus. According to a review of OPMD, prevalence of penetration or aspiration was up to 33% depending on the degree of swallowing difficulty.[2] In this patient, penetration was found after drinking a large amount of water and had impaired laryngeal elevation in a forward position. Therefore, he was trained to swallow with the Mendelsohn maneuver to keep the upper esophageal sphincter open, perform Shaker exercise to strengthen suprahyoid muscle, and keep chin tuck while swallowing to narrow the respiratory tract. In addition, dietary modification was prescribed as reported[2,10] because patients with OPMD and swallowing problems at the pharyngeal stage usually have weakness of pharyngeal constrictors and hypertonia of the upper esophageal sphincter.[9,10] For those with clear symptoms of cricopharyngeal dysfunction, botulinum toxin injection of 0-100 units with electromyographic guidance and under general anesthesia should be considered before cricopharyngeal myotomy.[10,11] The latter might cause complications such as paralysis of vocal cord (unilateral), dysphonia, or progression of dysphagia, and a recurrence of swallowing problem after surgery was reported.[11] If there is dysphagia in the oral stage such as drooling, a protocol of improving oro-motor function should be added.[3]

Concerning swallowing difficulty in this case, if there had been an early assessment, an appropriate advice and proper treatments, it could decrease malnutrition and other potential and harmful complications. Therefore, an initial treatment focusing on swallowing rehabilitation program should include modified foods, compensation strategies and swallowing exercise program, tailored to individual swallowing problems; and placing importance on safety eating with nutritional assessment at the same time.

In conclusion, patients with oculopharyngeal muscular dystrophy may present with swallowing difficulty due to weakness of pharyngeal constrictors and/or hypertonia of the upper esophageal sphincter. Pedigree and family history-taking is important to establish a pattern of transmission and diagnosis. Since there is no definite treatment at present, swallowing rehabilitation program is still very much needed and prescribed by rehabilitation physicians (physiatrists).

Declaration conflict of interest

The authors has no conflict of interest to declare.

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