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Esophageal Aperistalsis in a Patient with Lipoid Proteinosis

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ABSTRACT

Lipoid proteinosis is a rare disorder with autosomal recessive inheritance, characterized by progressive deposition of hyaline material in the skin, mucous membrane, and different organs of the body, resulting in a multitude of clinical manifestations. A 34-year-old woman presented with hoarseness, dysphagia, eyelid beeding, and acneiform scars on the facial skin and extremities. The patient was diagnosed clinically as having lipoid proteinosis, which was confirmed by laryngeal biopsy. The objective of the present report is to describe this rare entity. This case report also illustrates that lipoid proteinosis may show protean clinical features and yet may remain undiagnosed for many years.

KEYWORDS:
Acneiform scars, Eyelid beading, Hoarseness of voice, Hyaline material, Dysphagia

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INTRODUCTION

Lipoid proteinosis (LP) or Urbach–Wiethe disease is a rare, autosomal recessive disorder, which was first reported in 1929 by Erich Urbach and Camillo Wiethe.[1] This disorder was characterized by hoarseness from early infancy and various cutaneous manifestations, such as acneiform scarring, waxy papules, moniliform blepharosis, and non-cutaneous manifestations attributed to infiltration of hyaline-like material in the skin, larynx, and other organs.[2] The hyaline-like material is periodic-acid Schiff (PAS) positive and diastase resistant leading to the deposition of non-collagenous proteins and glycoprotein.[3]

To our knowledge, esophageal motility disorders have not been reported as a part of this disorder. We describe the first case of LP who presented with dysphagia caused by concomitant esophageal aperistalsis.

CASE REPORT

A 34-year-old Bakhtiari woman who was a known case of LP based on clinical and pathological diagnosis, referred to our hospital with hoarseness and dysphagia accompanied by cutaneous lesions. She presented with hoarseness and typical skin lesions since adolescence. About 7 years ago she developed a gradually progressive dys-

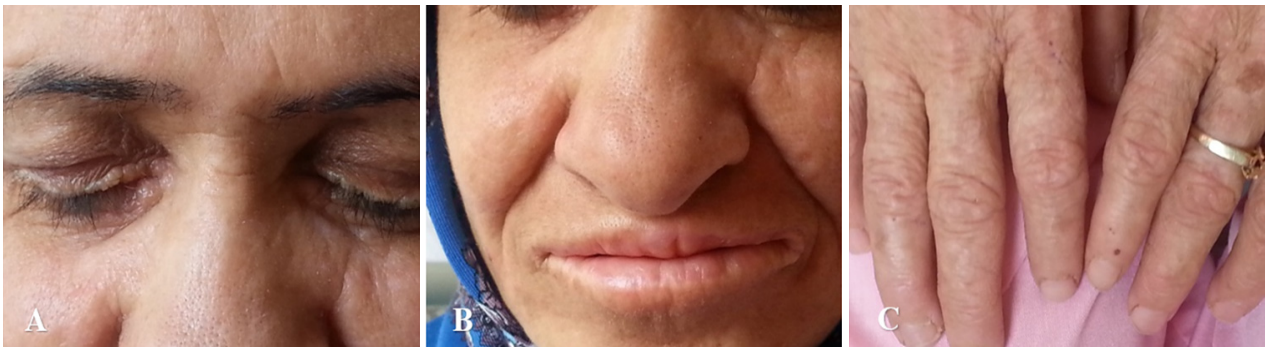

Fig.1: **Clinical findings in the patient with lipoid proteinosis who presented with dysphagia. A) eyelid beading (moniliform blepharosis); B) white infiltrates form lesions on the lips; C) multiple 2-3 mm, warty papules on the dorsum of the hand**

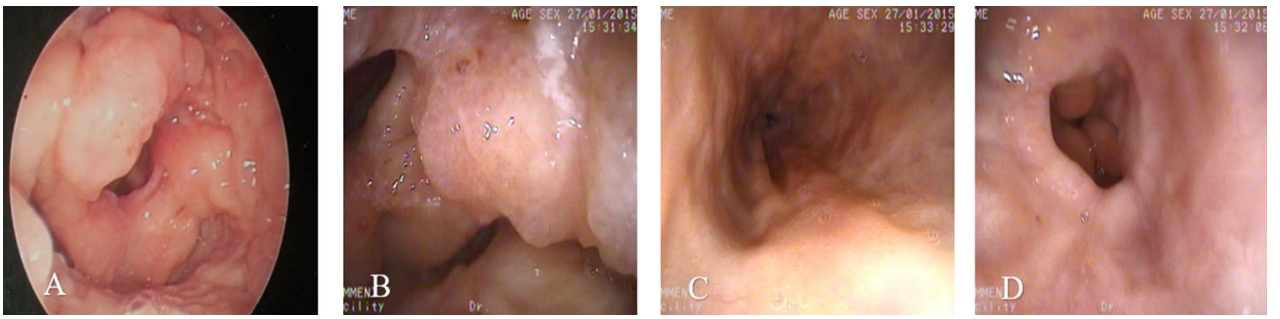

Fig.2: **Upper gastrointestinal endoscopic findings in the patient with lipoid proteinosis who presented with dysphagia. A) Larynx; B) Pharynx; C) Middle esophagus; D) Lower esophagus**

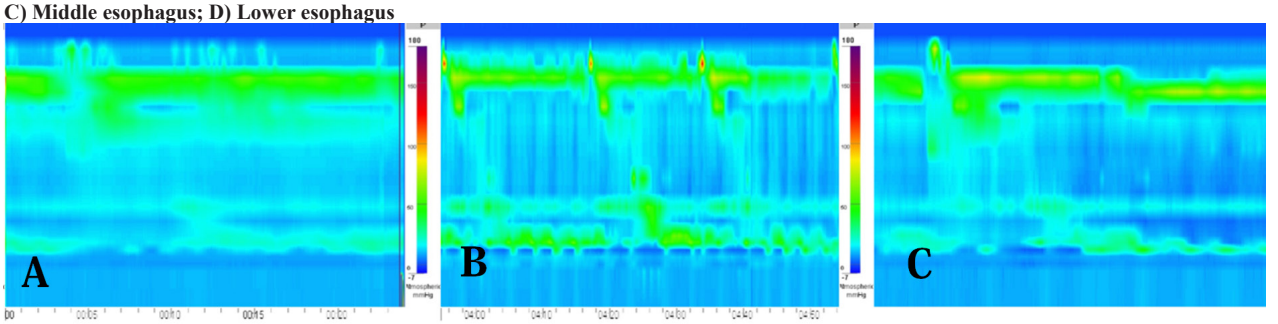

Fig.3: **High resolution manometric findings in the patient with lipoid proteinosis who presented with dysphagia. A) Average findings: DCI 31 mmHg.s.cm, peristaltic breaks 7.9 cm, distal latency 6.8 s, IRP 4 s 4.5 mmHg; B) Resting pressure (mean) 28.8 mmHg, resting pressure (minimal) 5.1 mmHg; C) A wet swallow**

phagia to solid food as a predominant symptom and complained of intermittent post-prandial chest pain without regurgitation or weight loss. She had no history of gastroesophageal reflux disease (GERD), seizures, visual disturbances, or respiratory obstruction. None of the other family members were affected. Examination revealed an otherwise healthy individual with typical clinical findings of LP (figure 1). Systemic examination and laboratory tests did not reveal any abnormalities. Upper gastrointestinal endoscopy showed widespread mucosal thickening and irregularity in the pharynx and larynx. Upper and middle parts of the esophagus were normal, but an esophageal ring with normal overlying mucosa was observed in distal esophagus (figure 2). High-resolution manometry showed absent peristalsis (figure 3).

DISCUSSION

LP or Urbach–Wiethe disease is an autosomal recessive genetic disorder, which is caused by mutations in chromosome 1q21 and the extracellular matrix protein 1 (ECM1) gene. The disease is diagnosed on the basis of clinical symptoms and histopathology. Affected indi-

viduals can be asymptomatic and carriers of the disease.[4] The accumulation of hyaline material in the dermis and the thickening of the basement membranes in the skin lead to the dermatological manifestation.[5] One of the typical dermatological signs of the disease is beaded papules on the eyelid, which was noted in our case.

Diffuse infiltration of the pharynx and larynx may cause respiratory distress, which requires tracheostomy but rarely LP is a life-threatening disease.[6] Our patient presented with hoarseness, typical skin lesions, and progressive dysphagia to solid food, but she had no respiratory symptoms. Some other common extracutaneous manifestations of the disease include epilepsy, mental retardation, and neuropsychiatric disorders.[4,7]

While gastrointestinal involvement is infrequent in LP, hyaline deposits have been shown in visceral biopsies and autopsy specimens from the esophagus, stomach, small bowel, and rectum.[8] In the largest case series of LP reported from turkey (including 14 and 10 cases), no esophageal involvement was reported.[9,10] Dysphagia has been described as a typical symptom of LP in two recent reports of cases with LP.[11,12] Infiltration of the upper third of the esophagus and a medium-sized sliding hiatal hernia were the only abnormal findings on upper endoscopy. We could only find one patients with LP in whom esophageal manometry was reported. Lima and colleagues reported a Brazilian patient with LP who was referred for the evaluation of epigastric pain, postprandial fullness, and bloating without esophageal symptoms.[13] Upper endoscopy showed multiple yellowish nodules throughout the esophagus, body of the stomach, and duodenum. Esophageal manometry was normal in this patient. An older report of a Chinese patient that dates back to 1988 [14] did not describe any esophageal symptoms, but the authors assessed the esophageal transit time in their patient before and after treatment with dimethyl sulfoxide. Interestingly, the mean esophageal transit time (normal value: < 10 seconds) decreased from 36.8 seconds before treatment to 5.8 seconds after 3 years of treatment. However, no manometric findings were available in the latter report.

Infiltration of hyaline material in the muscular layer of the esophagus may be the cause of the motility disorder found in our patient, but it was not possible to acquire a tissue specimen from the deep esophageal layers for histological diagnosis in our patient.

There is no specific and curative treatment for Urbach–Wiethe disease.[15] Current treatment includes oral steroids, dimethyl sulfoxide, D-Penicillamine, intralesional heparin, and etretinate, which are used for symptomatic treatment of disability and symptoms. Although acitretin was used there is no strong evidence on whether this drug is beneficial for the treatment of dysphagia. Some patients require tracheostomy due to the respiratory obstruction but it occurs rarely and life expectancy is usually normal .[16-18]

CONCLUSION

Beaded papules along the margin of eyelids, thickened protuberant lips, and hoarseness of voice, confirm the diagnosis of LP. Dysphagia as a clinical manifestation was not reported previously in LP but our patient presented with dysphagia and esophageal aperistalsis.

ETHICAL APPROVAL

There is nothing to be declared.

CONFLICT OF INTEREST

The authors declare no conflict of interest related to this work.

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