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Case Report / *Olgu Sunumu*

Lipoid proteinosis (Urbach-Wiethe disease): A rare entity and review of the literature

Lipoid proteinozis (Urbach-Wiethe hastalığı): Nadir bir durum ve literatürün incelenmesi

Murat Doğan[1], İbrahim Hıra[1], Burhan Balta[2], Ali Bayram[1], Cemil Mutlu[1]

[1]*Department of Otolaryngology, Health Sciences University, Kayseri Training and Research Hospital, Kayseri, Turkey*
[2]*Department of Medical Genetics, Health Sciences University, Kayseri Training and Research Hospital, Kayseri, Turkey*

ABSTRACT

Lipoid proteinosis (LP) is a rare disease. It may affect the skin, oral mucosa, pharynx, larynx and all visceral organs. In this article, we describe a 32-year-old female patient who applied with complaint of white swelling in mouth, limiting the movement of lip, and hoarseness. Papule-like itchy rashes particularly around the hands, elbows, and lips were observed in physical examination and bilateral blepharosis in eye examination. In videolaryngostroboscopy, thickened vocal cords and yellow papule formations covering the entire supraglottic region and oral mucosa drew attention. In punch biopsy samples taken from larynx and sublingual regions, hyperkeratosis-patterned multilayered epithelial and subepithelial amorphous hyaline material deposition was observed. In histochemical examination, positive staining with periodic acid-Schiff in basal membrane and negative staining with Congo red were obtained. We established a diagnosis of LP according to typical vocal cord involvement and histopathologic, genetic and clinical findings.

Keywords: Extracellular matrix protein 1; genetics; larynx; lipoid proteinosis; Urbach-Wiethe disease.

ÖZ

Lipoid proteinozis (LP) nadir görülen bir hastalıktır. Cilt, oral mukoza, farenks, larenks ve tüm iç organları etkileyebilir. Bu çalışmada ağız içerisinde dudak hareketini kısıtlayan beyaz renkli şişlik ve ses kısıklığı yakınması ile başvuran 32 yaşında bir kadın hasta sunuldu. Fizik muayenede özellikle el, dirsek ve dudaklar çevresinde papül benzeri döküntü ve göz muayenesinde iki taraflı blefaroz gözlendi. Videolarengostroboskopide, vokal kordlarda kalınlaşma ve tüm supraglottik bölgeyi ve oral mukozayı tutan sarı papül oluşumları dikkat çekti. Larenks ve sublingual bölgelerden alınan punch biyopsi örneklerinde, epitel ve subepitelyal yerleşimli hiperkeratoz paternli çok katmanlı amorf hiyalin materyal birikimi gözlendi. Histokimyasal incelemede, bazal membranda periyodik asit-Schiff ile pozitif ve Kongo kırmızısı ile negatif boyanma elde edildi. Tipik vokal kord tutulumu ve histopatolojik, genetik ve klinik bulgulara göre LP tanısı kondu.

Anahtar sözcükler: Ekstraselüler matriks protein 1; genetik; larenks; lipoid proteinozis; Urbach-Wiethe hastalığı.

Lipoid proteinosis (LP) has been first defined by Urbach and Wiethe in 1929 as "lipoidosis cutis et mucosae".[1] It is a rarely seen and recessively-inheriting hereditary disease.[2-4] Pathogenesis of LP is not known. Although it was named lipoidosis, it is thought that the hyaline material accumulating is a carbohydrate protein complex containing various levels of lipid.[5-9] Various studies have shown that LP might develop as a result of mutation in extracellular matrix protein 1 (ECM-1) gene in 1q21 chromosome.[9-11] Although it has been initially thought to be limited with skin and consequently oral mucosa and pharynx and larynx

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Correspondence: İbrahim Hıra, MD. SBÜ Kayseri Eğitim ve Araştırma Hastanesi Kulak Burun Boğaz Kliniği, 38010 Kocasinan, Kayseri, Turkey.
e-mail: dr.ibrahimhira@gmail.com

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