Two cases of recurrent oral ulcers with lipoid proteinosis

Lipoid proteinosisli iki olguda tekrarlayan oral ülserler

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Abstract

Lipoid proteinosis (LP) is a rare autosomal recessive disorder characterized by infiltration of amorphous hyaline material in the skin, oral cavity, larynx, and internal organs. Deposition may be in different parts of the body, especially in the skin, mucous membranes of the upper aero digestive tract, and internal organs. LP was diagnosed with clinical findings, family history and histopathological evaluation. LP may be very rarely associated with oral ulcers. In this report, we aim to describe two cases of patients affected by LP associated with painful and recurrent ulcerative lesions in the tongue.

Key Words: Lipoid proteinosis, oral ulcers, tongue

Özet

Lipoid proteinozis (LP) nadir görülen otosomal resesif bir genodermatoz olup deri, oral kavite, larinks ve iç organlarda amorf hyalin kristal materyal birikimi ile karakterizedir. Birikim özellikle deride olmak üzere, üst solunum-sindirim sistemi mukozası ve iç organlarda olur. Hastalarımıza klinik bulgular, aile öyküsü ve histopatolojik değerlendirme sonucu LP tanısı konuldu. Hastaların dilinde tekrarlayan ağrılı ülserler mevcut olup nadir rastlanılan bu vakaları LP oral ülser birlikteliği açısından sunmayı uygun bulduk.

Anahtar kelimeler: Lipoid proteinozis, oral ülser, dil

Introduction

Lipoid proteinosis is a rare autosomal recessive genodermatosis characterized by the defect of chromosome 1q21 which is on extracellular matrix protein gene 1. Skin, mucous membranes and internal organs are determined by PAS-positive hyaline material accumulation (1, 2). Clinical manifestation of LP begins in birth or early infancy with hoarseness, and/or failure cry soon after birth. Other conditions are yellowish white verrucous papules-nodules, acneiform scars, blisters, which may appear few years later (3, 4). Oral cavity is the most extensively affected area and oral manifestations include yellowish white infiltrate, thickened, enlarged tongue which impaired mobility. The others are dental agenesis, xerostomia and enlargement of the lips with fissuring (5). We found only 2 cases that developed ulcerated tongue associated with LP in the literature (6, 7). Therefore, we aimed to present these cases for contribution to the literature.

Case reports

Case 1

A 15-year-old woman was admitted to our clinic with recurrent painful ulcers of tongue. First of all recurrences were developed about 5 years ago. She had periods of recurrence and resolution with supportive treatment. She had a sister who also had LP but no history of oral ulcers. Dermatologic examination were hoarseness, dysphonia, thinning hair, hyperkeratosis of the feet, the size of pinhead percent of the upper eyelid, yellow-white papules and atrophic scars on face and hands. Oral examination revealed that the tongue was restricted of movement and stiffness, loss of tongue papillae and xerostomia. Uvula and tonsils were thickened and coarsened. Dental examinations were normal. The fissure at the lower lip and 4 ulcers were on the patient's tongue (Figure 1). Histological examination of skin biopsy material was taken from the tongue. It showed the deposition of pale, eosinophilic, hyaline material dermis, around blood vessels and adnexial structures were compatible with LP (Figure 2).

Case 2

A 5-year-old-boy without family history was admitted to our clinic with hoarseness and acneiform scars of the hand and face. Symptoms with very painful ulcers, especially on the tongue, that preventing eating had begun 1 year ago. No specific treatment had been given, and ulcers had been spontaneously resolving within few days. The diagnosis of LP is made based on clinical findings, family history and confirmation of histopathological evaluation. The second case had same

dermatological findings, in addition to this; there were also 1 ulcer on the ventral surface and the fissures at the angle of the mouth (Figure 3). Histological examination of skin biopsy material was taken from tongue and showed the similar findings with regard to first case. The other laboratory findings of two patients were normal. Both patients were photographed by taking the written informed consent from the patients' families.

Discussion

Lipoid proteinosis is a rare benign and slow progressing autosomal recessive inherited genodermatosis. Histological examination reveals widespread deposition of hyaline-like material and distruption of basement membrane around blood vessels and at the dermal-epidermal junction. Extracellular matrix protein 1 gene has been implicated (3). Accused of this gene produce a protein that abnormally accumulates caused the increased thickening of the vocal cords and is thought to be causing vocal cord paralysis and hoarseness. Over time, increased mucosal infiltration can cause upper respiratory tract infection, and sometimes the need tracheostomy (8). In addition, episodic inflammation of the parotid gland and submandibular gland, poor oral hygiene, frenilumun thickening can be seen due to the shortness of the tongue (3, 4).

In our cases, hoarseness, dysphonia, hair thinning, hyperkeratosis, yellow-white papules the size of pin head, atrophic scars on face and hands, papillae loss on tongue and limitation of movement, thickening of the uvula and tonsils were observed which similar to literature. Recurrent painful tongue ulcers and lip fissures were ın both cases for a few years, and spontaneously resolutions within days were seen. We found 2 cases of coexistence LP with recurrent ulcers in the mouth in the literature. 3-year-old patient with severe oral ulcers, which started a month ago and the other patient was 27-year-old had symptoms from childhood. In our cases tongue ulcers were continuing for 5 and 1 year. Although previous articles have not described the mechanism of this association; xerostomia, episodic inflammation of the glands in the oral mucosa, poor oral hygiene, thickening of frenulum, shortness of language impaired the integrity of the oral mucosa. So, recurrent ulcers may occur in mouth especially in the tongue. As a result, we presented the cases of LP with oral ulcers that this lesions are extremely rarely seen.

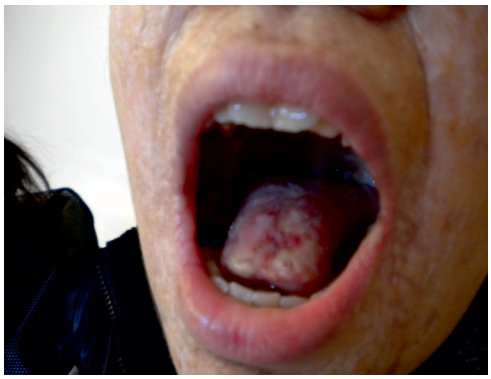

Figure 1: Figure demonstrating the fissure at the lower lip and 4 ulcers on the tongue

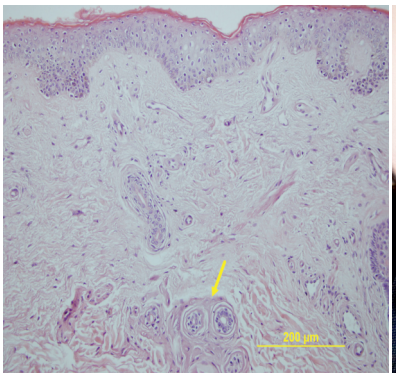

Figure 2: Marked skin patches and hyalinization of vascular structures around the papillary dermis (H&E x 10).

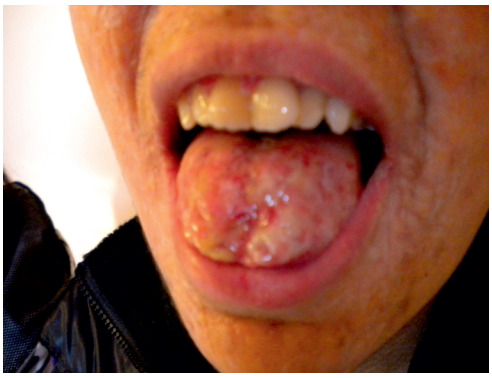

Figure 3: Figure demonstrating 1 ulcer on the ventral surface of the tongue and the fissures at the angle of the mouth.

Yazarlarla ilgili bildirilmesi gereken konular (Conflict of interest statement) : Yok (None)

References

1. Touart DM, Sau P. Cutaneous deposition diseases. Part I. J Am Acad Dermatol 1998; 39: 149–71
2. Chan I, Liu L, Hamada T, Sethuraman G, McGrath JA. The molecular basis of lipoid proteinosis: mutations in extracellular matrix protein 1.Exp Dermatol 2007; 16: 881–90
3. Aroni K, Lazaris AC, Papadimitriou K, Paraskevakou H, Davaris PS. Lipoid proteinosis of the oral mucosa: case report and review of the literature. Pathol Res Pract 1998; 194: 855–9
4. Disdier P, Harlé JR, Andrac L, Swiader L,Weiller PJ. Specific xerostomia during Urbach–Wiethe disease. Dermatology 1994; 188: 50–1
5. Bazopoulou-Kyrkanidou E, Tosios KI, Zabelis G, Charalampopoulou S, Papanicolaou SI. Hyalinosis cutis et mucosae: gingival involvement. J Oral Pathol Med 1998; 27: 233-7
6. Sargenti Neto S, Batista JD, Durighetto AF Jr. A case of oral recurrent ulcerative lesions in a patient with lipoid proteinosis (Urbach-Wiethe disease). Br J Oral Maxillofac Surg 2010; 48: 654-5
7. Kaya TI, Gunduz O, Kokturk A, Tursen U, Ikizoglu G. A life-threatening exacerbation of lipoid proteinosis. J Eur Acad Dermatol Venereol 2002; 16: 286–8
8. Ramsey ML, Tschen JA, Wolf JE Jr. Lipoid proteinosis. Int J Dermatol 1985; 24: 230-2.