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Short communication

A case of oral recurrent ulcerative lesions in a patient with lipoid proteinosis (Urbach–Wiethe disease)

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Abstract

Lipoid proteinosis is a rare autosomal recessive genodermatosis characterized by deposition of amorphous hyaline material in different parts of the body, especially the skin, mucous membranes of the upper aerodigestive tract, and internal organs. A clinical manifestation of LP usually begins as a hoarseness and failure cry soon after birth or in the first years of life. However, other conditions may occasionally appear few years later. Oral cavity is most extensively affected area by the disease and the main oral abnormalities include diffusive infiltration of white pea-size plaques and stiffening of the tongue as well as inability to protrude it. In this report, we describe the case of a woman affected by LP presenting recurrent ulcerative lesions in mouth associated with xerostomia.

Keywords: Urbach–Wiethe disease; Lipoid proteinosis; Xerostomia; Oral ulcers; Tongue

Introduction

Lipoid proteinosis is a rare autosomal recessive genodermatosis characterised by deposits of amorphous hyaline material mainly in the skin and mucous membranes.[1,2] We describe a woman who presented with recurrent ulcerative lesions in her mouth associated with xerostomia.

Case report

A 27-year-old woman was admitted complaining of ulcerating lesions of her tongue, the first of which had developed during childhood; she had had periods of recurrence and spontaneous resolution. Lipoid proteinosis had been diagnosed then by a dermatologist. She had a brother who also had lipoid proteinosis and who had similar lesions in his mouth. On examination she was hoarse, and had alopecia, blepharosis, hyperkeratosis of the hands, and scars on her skin (Fig. 1). Intraoral examination showed a stiff tongue with little mobility and no papillae on the dorsum (Fig. 2). There were three painful ulcerating lesions on the ventral surface (Fig. 3). In addition, her lips were partially everted with fissures at the angle of the mouth. The uvula and palatine tonsils were thickened and rough (Fig. 2). Xerostomia was also evident (Figs. 2 and 3).

Agenesis of teeth 12 and 22 was noted radiographically. The laboratory findings were within the reference ranges. Histopathological examination of the skin biopsy specimen obtained at first diagnosis was reviewed, and a diagnosis of aphthous stomatitis with xerostomia was suggested. She was given no specific treatment for the ulcers, though she was advised to use artificial saliva to lubricate her mouth for the foreseeable future. After a year of follow-up she has reported no further signs or symptoms.

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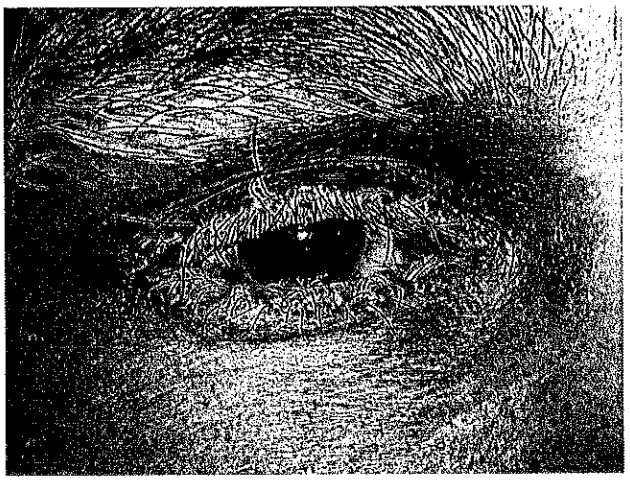

Fig. 1. Eye with beaded papules on the eyelid (moliniform blepharosis).

Fig. 2. Note the absence of papillae on the dorsum of the tongue and the presence of everted lips and fissures at the angle of mouth. Uvula and palatine tonsils are thickened and rough.

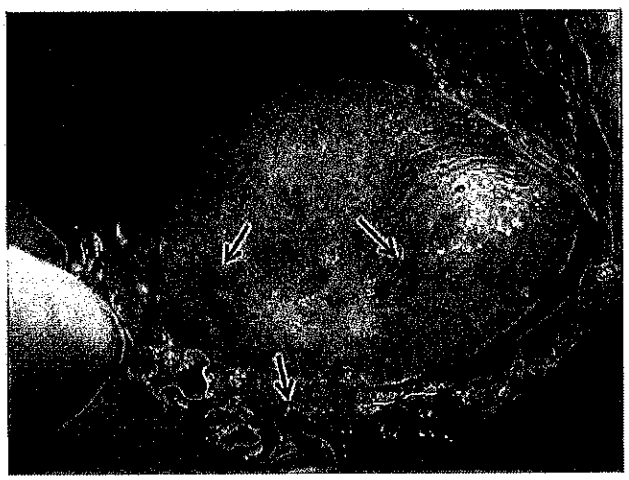

Fig. 3. The reddish ulcerated lesions on the ventral surface of the tongue measuring 1.0 cm (arrows). Also it could be evidenced the reduced mobility caused by a thickened sub-lingual frenulum.

Discussion

Lipoid proteinosis usually follows a benign and slow progressive course.[3] It affects both sexes equally and the incidence is higher in Sweden and South Africa than elsewhere.[3,4] The extracellular matrix protein 1 (*EMC1*) gene was recently named as the responsible gene, but its exact biological functions have not been elucidated.[2,5] Patients develop hoarseness and failure to cry soon after birth, which is the first sign.[4,6] Other signs include beaded papules on the eyelids, skin eruptions, hyperkeratosis of the hands and elbows, and alopecia.[1,3] Features may develop in the oral cavity in childhood, or not all. The hallmarks are stiffening of the tongue and inability to protrude it, thickened and roughened uvula and palatine tonsils, and yellowish-white infiltrates in the form of small papules. Others uncommon conditions are gingival overgrowth, dental agenesis, and xerostomia.[3,7]

Histologically, it presents as deposits of amorphous, extracellular eosinophilic hyaline material (it stains for periodic acid Schiff and is resistant to amylase) in the upper thickened connective tissue; in the lower part changes are focal with a hyaline mantle around the vessels. The diagnosis is based on clinical findings and confirmation by histopathological examination.[1,3]

Our case is typical of lipoid proteinosis, except that our patient had continued to develop oral ulcers since childhood. Only a few reports have mentioned the involvement of salivary glands, and the consequences have been described in only a few reports[3,7]; we found only one case of a girl who developed an ulcerated tongue.[4] The course of the disease and the therapeutic possibilities are still being debated.[1,3–5]

Conflict of interest

There is no conflict of interest.

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