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A Rare Genodermatosis: Lipoid Proteinosis

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Abstract

Lipoid proteinosis is a rare genodermatosis that manifests with hoarseness and cutaneous findings. Its skin manifestations start with vesicles during the neonatal period, later scar formation and lichenification are observed. Beaded papules on the eyelids, which is referred to as "moniliform bleapharosis" is a typical finding. Here, we present a case of this rare disease with the typical manifestations of the disease.

Keywords: Deposition, hoarseness, lipoid proteinosis

Nadir Bir Genodermatoz: Lipoid Proteinozis

Öz

Lipoid Proteinoz nadir görülen bir genodermatozdur. Hastalık ses kısıklığı ve deri bulguları ile kendini gösterir. Yenidoğan döneminde vezikül formasyonu ile başlayan bulgular ilerleyen zamanlarda sikatris oluşumuna ve likenifikasyona yol açar. Kirpik kenarında papüllerin görülmesi oldukça tipik olup "moniliform blefarozis" olarak adlandırılır. Bu nadir genodermatozun tipik bulgularını taşıyan bir olguyu paylaşmak istedik.

Anahtar Kelimeler: Birikme, ses kısıklığı, lipoid proteinozis

Lipoid proteinosis is a rare genodermatosis that manifests with hoarseness and cutaneous findings. The deposition of hyaline material within the basement membrane of the blood vessels and the dermoepidermal junction is the key histological finding that leads to the characteristic signs and symptoms of the disease.[1,2] Here, we present a case of lipoid proteinosis that manifests the typical findings of the disease.

Case Presentation

A 33-year-old male patient approached the dermatology outpatient clinic with the primary complaint of bleeding and burning in the inguinal region. The patient has visited several dermatology clinics complaining of recurrent lesions at different locations since he was born. The dermatologic examination revealed macerated plaques in the inguinal fold, verrucous plaques on the elbows (Figure 1a), ice-pick atrophic scars in the malar region within the beard, verrucous papules in the submental area (Figure 1b), small yellowish-white papules on the eyelid margins (Figure 1c), and yellow pearly papules around and between the eyebrows (Figure 1d). The examination of the oral mucosa revealed macroglossia. The dermoscopic examination of the elbow plaques and eyebrow papules revealed light-red to light-brown blotches, which were conspicuous of deposition of hyaline (Figure 2). The examination of other systems was unremarkable except for hoarseness, which the patient recalls has started during childhood. The past medical history revealed spinal radiculomyelopathy, which was under medical control. Family history revealed a cousin with similar skin findings, who also was undiagnosed.

An incisional biopsy, with the pre-diagnosis of lipoid proteinosis, was performed from the left elbow plaque by general surgery. The histopathology revealed amorphous eosinophilic material distributed interstitially, periadnexially, and perivascularly in the dermis. Periodic-Acid-Schiff staining was positive. A histopathological diagnosis of lipoid proteinosis was given.

The blood chemistry was unremarkable except for an LDL cholesterol of 104 mg/dL (N < 100) and HDL cholesterol of 37 mg/dL (N > 40).

The patient was referred to otolaryngology for his hoarseness. The endoscopic examination revealed that the entire oropharyngeal and laryngeal mucosa were thick and dry starting from the vermillion border, and the plications were less pronounced and epithelized at certain parts. The epiglottis was omega-shaped, thick, and less mobile. Thickening and dullness were pronounced in the vocal cords. The endoscopic and microscopic findings were consistent with the diagnosis of lipoid proteinosis.

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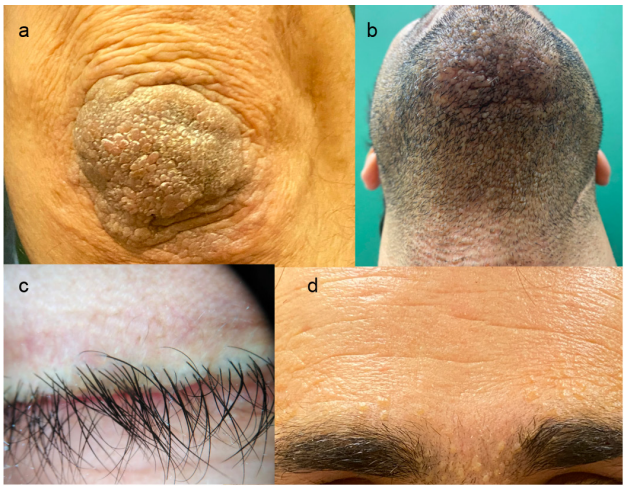

Figure 1. a–d. Typical cutaneous findings of the disease. (a): skin-colored lichenified plaques with overlying verrucous papules; (b): papules and acneiform scar formation; (c): moniliform blepharosis; (d): forehead papules.

Acitretin (20 mg/day) treatment was initiated. Occlusive treatment with 40% salicylic acid (within vaseline) was applied to the keratotic plaques on elbows.

Discussion

Lipoid proteinosis, which is also referred to as the Urbach–Wiethe disease, is a rare autosomal recessive disorder that occurs due to a loss of function mutation in the extracellular matrix protein 1 gene (ECM-1).[1-3] The mutation of the ECM-1 gene leads to the deposition of hyaline material in the skin and mucous membranes along with the internal organs.[3]

The usual presenting symptom of the disease is hoarseness, which occurs due to hyaline deposition in the mucosa of the upper respiratory tract. The cutaneous manifestations start with vesicles in the neonatal period. During childhood, minor trauma or friction may easily damage the skin leading to blistering and acneiform scar formation on the face and the extremities.[1,3] With time, the skin thickens, especially on the face and the extremities. Skin-colored lichenified plaques with overlying verrucous papules become evident at frictional areas, such as the knees, elbows, axillae, and inguinal folds.[3,4] Similar to the previously reported cases, the physical examination of our patient also revealed these typical plaques on the elbows, inguinal folds, and submental regions. Indeed, the primary complaint of the patient was the ulceration of such a plaque in the inguinal fold. Another striking feature of this disease is the beaded papules on the upper eyelid margin, which is referred to as "moniliform blepharosis" in the literature. Skin-colored papules and atrophic scars on the forehead are also typical of the disease.[2-5] Moniliform blepharosis (Figure 1c) and forehead papules (Figure 1d) were also prominent in our patient. Woody induration of the tongue along with indentations may also occur due to hyaline deposition, leading to macroglossia, which we also observed in our patient.[2,3] Neuropsychiatric abnormalities and seizures may be observed due to the deposition in the amygdala.[5]

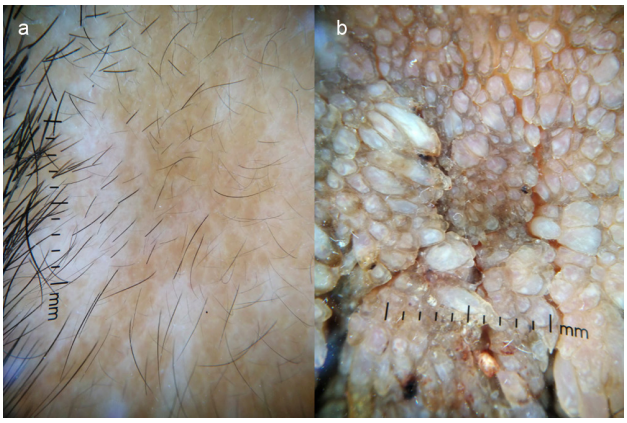

Figure 2. Dermoscopic examination of the lesions revealing the hyaline deposition.

Unfortunately, the treatment of the disease has not been evaluated in large series since this is a rare disorder, and the level of evidence is of case reports.[6] Acitretin, at a daily dose of 0.5 mg/kg for 6 months, is an effective treatment for lipoid proteinosis.[7] D-penicillamine, 600 mg/day for 2 years, has been reported to be an alternative treatment.[8] Furthermore, laser resurfacing with 2940 nm ablative Er:YAG laser can be used for thick verrucous plaques, achieving cosmetically acceptable results.[9]

Lipoid proteinosis is a rare genodermatosis that impacts the quality of life of the patients dramatically since it manifests with hoarseness and disfigurement due to mucocutaneous accumulation of hyaline material. A definitive treatment modality has not been determined due to the rarity of the disease.

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