Dermatology Practical & Conceptual

Association Between Lipoid Proteinosis and Coeliac Disease

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Lipoid proteinosis, also known as Urbach-Wiethe disease, is a rare autosomal recessive condition, characterized by a mutation of extracellular matrix protein 1(ECM1), leading to deposition of collagenous material in the skin and tissues [1]. This disease may be associated with diabetes and other abnormalities. We report a case of lipoid proteinosis associated with celiac disease in a 23-year-old man.

A 23-year-old patient was admitted to our department for facial edema evolving for 1 year, with no other associated signs. The dermatological examination revealed an infiltrated edema of the face, predominantly on the forehead, associated with discrete 3 mm yellow papules in the ear pavilion (figure 1), without mucosal involvement. The histological study of the papule showed a homogeneous, eosinophilic, PAS-stained dermal deposit of hyaline substance, in favor of lipoid proteinosis. Laboratory tests revealed hypochromic microcytic anemia (hemoglobin of 9 g/dl), ferritin 7ng/ml, hypocholesterolemia 0.7 mmol/l with normal albumin and protein. The patient was further evaluated with an upper endoscopy. The histological examination of the duodenal biopsy revealed atrophic pangastritis with villous atrophy. Test IgA anti-transglutaminases were highly positive at low titer. The diagnosis of coeliac disease was established. The workup for other disease locations of lipoid proteinosis was negative. The therapeutic approach was to put the patient on a gluten-free diet and acitretin for his skin lesions. After 3 months of treatment with acitretin, the dermatological examination noted a minor improvement of the edema. The cutaneous lesions were still present, without new lesions. Then, he was lost to follow-up.

Lipoid proteinosis, also called hyalinosis cutis mucosae, is a rare, non-life-threatening condition and was first described in 1929 by a dermatologist and an otorhinolaryngologist, E. Urbach and C. Weithe [2]. Around 300 cases have been reported in the literature with higher prevalence in South Africa and Sweden.

Hoarseness of the voice is usually the first sign of this disease that appears in childhood. The dermatological manifestations include vesiculobullous lesions, hyperkeratotic plaques on the extensor surfaces of elbows, knees, and hands, yellow papules, and multiple acneiform and pox-like scars that are seen on the face [3]. Moniliform blepharitis is the

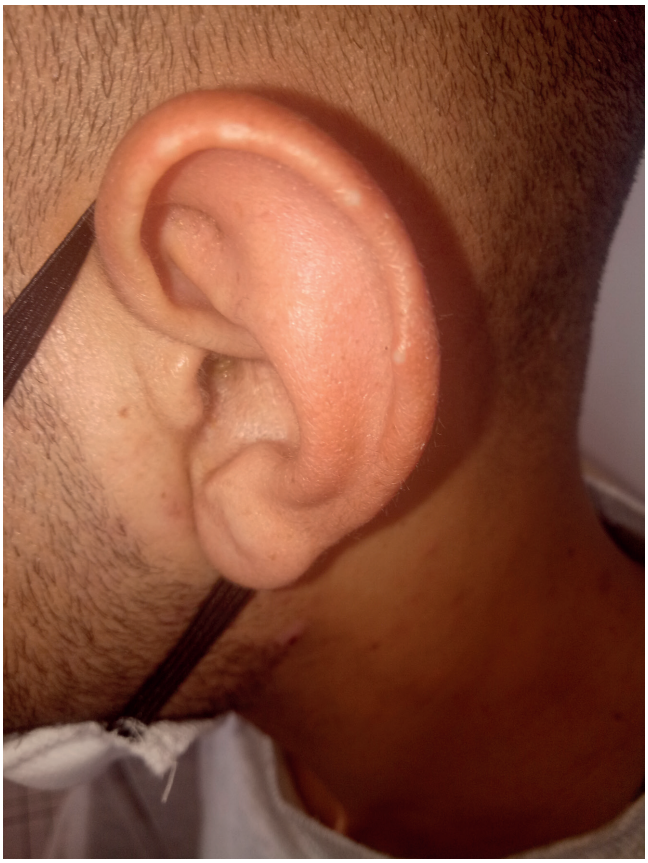

Figure 1. Yellowish-white papules of the ear.

characteristic ocular involvement, affecting the eyelid margins. This disease may affect all organ systems of the body.

Histologic features consist of thick homogeneous, eosinophilic hyaline material, periodic acid- Schiff positive, in the basement membrane and the dermis [4].

Some cases in the literature have mentioned the association of lipoid proteinosis with diabetes [5], and with epilepsy [6]. This is the first report describing an association with coeliac disease.

Currently, there is no effective treatment. Several molecules have been used with different results, such as D-penicillamine, dimethyl-sulfoxide, and retinoids for their inhibitory effect on collagen. Also, CO2 laser, dermabrasion, and chemical peeling represent other therapeutic means for the management of cutaneous lesions [3].

Knowledge of this rare disease will aid health professionals in diagnosing it early, and in providing the appropriate treatment to improve the quality of life.

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