CASE REPORT

Lipoid Proteinosis: case report and review
Khalid Mohammed Alshahrani, MD

Consultant Dermatologist, Department of dermatology, King Khalid University
Abha, Kingdom of Saudi Arabia

ABSTRACT
Lipoid proteinosis (Urbach-Wiethe disease) is a rare, recessively inherited disorder that is characterized by the deposition of hyaline-like material in the skin, oral cavity, and other tissues. It usually appears in infancy with hoarseness.

KEY WORDS: Lipoid Proteinosis, hyaline-like material

INTRODUCTION
Lipoid proteinosis (LP) is a rare disorder that is inherited as an autosomal recessive trait and has no sexual preference. LP was probably first described by Siebenmann in 1908 as a case of hyperkeratotic skin and yellow-white infiltrates of the oral mucosa.

In 1929 Urbach and Wiethe established the disorder as a clinical and histologic entity under the title “lipoidosis cutis et mucosae.” In 1932 Urbach changed the terminology to “lipoid proteinosis” because his histochemical studies revealed that the substance deposited in the skin had staining characteristics of both lipid and protein. Because the basic biochemical or chromosomal defect in LP is unknown, the use of the eponym “Urbach-Wiethe disease” has been suggested. We report a case of LP and review the literature.

CASE REPORT
A 16-year-old Saudi boy (first seen by Dr. Khalid alshahrani) came complaining of lesions on his face that resembled acne scars. The patient was the product of a full-term gestation and was born to non-consanguineous parents. At birth his skin appeared normal; the lesions developed during the first few years of life.

His parents also reported that he had history of hoarseness since birth, that gradually progressed to improve as he grew up. They denied any episodes of dyspnea. The patient has 5 other siblings, 4 boys and one girl and one sibling had died at the age of 5 years, and the cause of death could not be ascertained. One of his uncle was known to have similar symptoms. He did not have any other medical illness.

Examination
Revealed multiple scattered, pocklike, atrophic scars on the face (Fig. 1), neck, lateral aspects of the chest, and inner surfaces of the forearms. Diffuse, firm thickening of the skin of the extremities with hyperkeratosis on the elbows, knees, and the back of the hands was noted. Clusters of small papules were seen in the axillae. A row of skin-colored, coalescing papules appeared on the upper and lower eyelid.

Correspondence: Dr. Khalid Mohammed Alshahrani, Consultant Dermatologist, Medical City, King Khalid University, Saudi Arabia
Tel.: +966553366211, +96655563001. E.mail: khaliderma@gmail.com - al-salim@hotmail.com

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margins. The eyebrows were normal but the eyelashes were scanty. Intraoral examination showed irregular, submucosal infiltrates on the soft palate, and a moderately enlarged tongue that was bound to the floor of the mouth (Fig. 2). The tongue was firm and inelastic. And he had limited protrudtion of the tongue. Laryngeal examination done by ENT specialist, revealed similar plaques on the tonsils, the posterior wall of the pharynx, the epiglottis, and the aryepiglottic folds. The vocal cords appeared grey and thickened but without nodules. Neurologic examination was normal. The patient’s general level of intellect and language skills were normal. Results of the ophthalmologic examination were normal.

HISTOPATHOLOGY

The biopsy taken from side of face, in the epidermis showed orthokeratosis, papillomatosis, acanthosis and homogenous eosinophilic hyalanized material in the papillary dermis and around blood capillaries (Fig. 4, 5). PAS stain showed positive reaction and homogenous eosinophilic hyalanized material (Fig. 6, 7).

Fig. 1 A Lipoid proteinosis. Note diffuse infiltration, extensive facial scarring, B Closeup of the forehead lesions.

Fig. 2 Oral examination.

Fig. 3 Eyelid examination showed tiny papules on eyelids and around orbital.

Fig. 4 Orthokeratosis, papillomatosis, acanthosis and homogenous eosinophilic hyalanized material in the papillary dermis and around blood capillaries.
DISCUSSION

More than 300 cases of LP that have predominantly involved persons of European ancestry have been reported. Although, some of the patients who exhibited light sensitivity probably had erythropoetic protoporphyria, and were wrongly diagnosed. Large kindred’s have been reported from South Africa, where most of the cases can be traced to a German immigrant.\textsuperscript{6,7} LP is an autosomal recessive disorder.\textsuperscript{8,9} Heterozygous carriers are virtually normal but may possibly be identifiable by minor signs such as abnormalities in tooth formation.\textsuperscript{10} Our patient meets the clinical, histopathologic, and ultrastructural criteria for the diagnosis of LP. Patients with this genodermatosis can usually be recognized instantly because of their course voice, inability to protrude the tongue, and thickened eyelids. The last sign, so-called “moniliform blepharosis,” is a particularly useful clue for the diagnosis of LP. It results from involvement of the eyelid margins with tiny papules, which produce the typical “string of beads” appearance (Fig. 3). Further examination usually reveals plaques that appear to be verrucous or xanthomatous in areas of trauma, particularly on the knees, elbows, hands, and feet. Involvement of the scalp often leads to loss of hair. Nail growth and dental development may be hampered. Hypoplasia or aplasia of the permanent maxillary lateral incisors and premolars is most frequently noted. The lips may be everted and their surfaces studded with tiny yellow papules. Over time the entire skin takes on a yellow, waxy appearance with diffuse thickening, particularly in the flexures. Mucosal signs are present at birth or appear within the first few years of life. The inability to cry at birth indicates early laryngeal involvement, but this rarely leads to respiratory obstruction. Characteristic deposits have been found in virtually every organ of the body including the central nervous system. The appearance of bilateral “bean-shaped” calcifications in the region of the anteromential temporal lobes is pathognomonic and is sometimes responsible for
epilepsy or behavioral defects. The absence of neurologic manifestations in our patient probably relates to his young age.

The characteristic histologic findings in both skin and mucosae include extensive deposits of amorphous eosinophilic material in the papillary dermis, along capillaries, and around sweat coils. This hyaline-like material has a strong reaction to the periodic acid-Schiff stain, which most likely indicates the presence of glycoproteins. Positive reactions to lipid stains such as Sudan black, that identify neutral fats are also seen. This probably results from the affinity of lipids for glycoproteins, and not from a primary metabolic defect in lipid metabolism. No reaction is usually seen to stains for amyloid, such as crystal violet. Electron microscopic findings show islands of fine granular material in the papillary dermis and around endothelial cells, pericytes, and smooth muscle cells of the blood vessels. In addition, concentric calcified granules and multiplication of the basal lamina around blood vessels or the dermoepidermal junction has also been described. The pathogenesis of LP is unknown. LP appears to share certain characteristics with the Lysosomal storage group of diseases, which makes it possible to explain the increased amount of glycoprotein as a result of single or multiple enzyme defects. Strong evidence also indicates that alterations in the distribution of genetically distinct collagen types may be primarily responsible for the deposition of the hyaline-like material. No known therapy exists for LP. Removal of vocal cord infiltrates may provide temporary relief from hoarseness. There is a report of one patient treated with dimethyl sulfoxide with excellent results.

REFERENCES