

Lipoid Proteinosis: Curious Case of Two Siblings!

Sir,

A 40-year-old male patient born out of a non-consanguineous marriage presented to us with a 25 year history of hoarseness of voice with thickening of skin over face with longitudinal and transverse folds with multiple skin colored asymptomatic thick lesions over both elbows, knees, axillae, groin, and upper eyelids. He also gave history of recurrent painful pus filled skin lesions. Similar complaints were also present in his 33 years old younger brother. On examination, both siblings had well-defined, bilaterally symmetrical skin to yellow colored verrucous papules and plaques over elbows, knees, and axillae [Figure 1a-d]. Multiple hyperkeratotic waxy beaded papules were present over margins of both upper eyelids and over angle of mouth [Figure 2a]. They had thickened facial skin over forehead with increased longitudinal and transverse skin folds as well radiating linear scars around the mouth [Figure 2b]. Both brothers had yellowish papules and lichenification over the scrotum. The tongue was enlarged with restricted movements in the younger brother. Multiple discrete, round to oval pock like scars were present over the trunk in both brothers.

Hematological and biochemistry profile of both patients were normal. A punch skin biopsy was sent from the most characteristic lesion. In H and E stain, all specimens showed similar findings which included acanthosis and papillomatosis in the epidermis. Dermis was thickened with large deposits of extracellular hyaline material in upper dermis and perivascular areas [Figure 3]. Lipoid proteinosis is a rare autosomal recessive disorder, characterized by infiltration of hyaline material into the skin, oral cavity, larynx, and internal organs.^[1] The disorder is caused by homozygous or

compound heterozygous mutation in the extracellular matrix protein 1 (ECM1).^[2]

It may present with a weak cry at birth, but usually presents later with hoarseness of voice within the first few years of life which gradually progresses as the age increases.^[3] Due to infiltrates on the undersurface of the tongue, there may be restriction of tongue movements. The first skin lesions are often blisters in early childhood, which become eroded and crusted after minor trauma.^[4] The skin of patients with lipoid proteinosis is highly susceptible to damage by appearance of chickenpox like scars and yellowish papules. Infiltration of skin then presents with groups of warty plaques on axilla and elbows. Beaded papules on the eyelid margins (moniliform blepharosis) are a characteristic finding in about two-third of patients. There may be loss of eyelashes or patchy alopecia due to scalp involvement.^[5] Histologically, the epidermis shows acanthosis and irregular acanthosis. The dermis is thickened and



Figure 1: Bilaterally symmetrical verrucous papules and plaques over elbows (a and b) and axillae (c and d)

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Figure 2: Thickened facial skin with increased longitudinal and transverse skin folds along with beaded papules over upper eyelids (a) with waxy lesions over angle of mouth (b)

the upper dermis contains large deposits of periodic acid–Schiff (PAS) positive extracellular hyaline material. There is also thickening of the (PAS-positive) basement membranes at the dermal–epidermal junction and around the blood vessels and sweat glands. The prognosis is generally good and disease progression is only until early adulthood. Treatment modalities reported in the literature include oral steroids, dimethyl sulphoxide, intralesional heparin, and tretinoin.^[6]

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

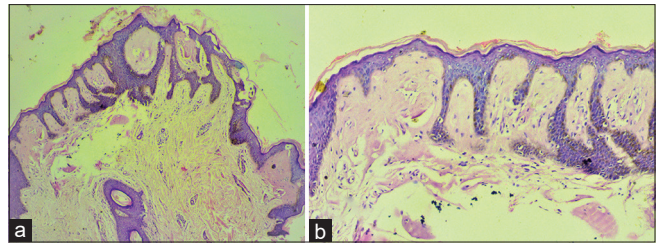


Figure 3: Papillomatosis with large deposits of extracellular hyaline material in the upper dermis (H and E a-40×, b-100×)

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Conflicts of interest

There are no conflicts of interest.

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