Hoarseness of Voice and Skin Lesions Since Childhood

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17-year-old male from West Bengal presented with history of recurrent blisters over sites of trauma and decreased vision and a hoarse voice since childhood. He was born of a non-consanguineous marriage and one of his siblings had similar skin lesions but to a lesser extent.

On examination, he had waxy infiltrated papules along the eyelid margins, nose, pinnae and axillae. There were pock-like scars, open and closed comedones seen over the face (Figure 1), warty papules and plaques over the gluteal cleft and elbows (Figure 2). Atrophic scars with mottled pigmentation were seen over the entire back and lower limbs. Cicatricial alopecia was seen over the scalp. He had difficulty in protrusion of tongue (Figure 3) and his buccal mucosa was infiltrated and thickened. He had decreased visual acuity (2/60) with bilateral superior lens subluxation with phacodonesis and iridodonesis. Histopathology of skin showed deposition of homogenous eosinophilic material which was PAS positive and diastase resistant in the superficial dermis and around capillaries and also around sweat ducts (Figure 4) in the dermis and subcutis.

Laboratory investigations including complete blood counts, renal and liver function tests, lipid profile, blood sugars, serum electrophoresis and urine porphyrins were within normal limits.

Diagnosis: Lipoid Proteinosis

Lipoid proteinosis is a rare autosomal recessive disorder due to loss of function mutations in the gene encoding extracellular matrix protein 1 (ECM1) on band 1q21.1 The ECM1 gene product is a glycoprotein with functional roles in skin physiology and homeostasis. There is progressive deposition of hyaline-like material which is PAS positive and diastase resistant in skin, mucosa, larynx and internal organs.

The first clinical sign of the disease is often a weak cry or hoarseness. Vesicles and haemorrhagic crusts are seen following trauma which resolve with scarring. The skin becomes diffusely thickened and waxy and develops a yellow discoloration. Beaded arrangement of waxy papules along the lid margin- moniliform blepharosis is characteristic.2 Verrucous lesions may be seen over the elbows, knees, dorsa of hands and the gluteal region. Scalp lesions may cause patchy alopecia. Tongue is usually firm and its mobility may be limited due to infiltration of the frenulum. The tonsils and the oropharynx may be infiltrated. Recurrent parotitis and dental anomalies can occur. Infiltration of the eyelids induces malpositioning of the eyelashes, causing corneal ulceration. Alopecia of the eyelashes and eyebrows can occur. Lens subluxation has been reported.3 Neurologic manifestations include seizures, memory deficits and abnormal behaviour. Bilateral, intracranial bean shaped suprasellar calcification may be seen. The respiratory tract may be involved by infiltration of the vocal cords and obstruction. The abnormal deposition of hyaline material has been detected histopathologically in many internal organs, although this is typically asymptomatic. Lipoid proteinosis is compatible with a normal life span except for the risk of respiratory obstruction in infants. The clinical differential diagnosis includes erythropoietic protoporphyria, amyloidosis, papular mucinoses and xanthomatosis. Treatment of this condition is usually unsatisfactory.

References

