Case 7

A 9 years old Thai girl from Bangkok

Chief complaint A hoarseness of voice since she were born, multiple yellowish papules on the rim of both upper lids for 3-4 years. There was no other skin lesions elsewhere.

Past history She had no other history of underlying disease or drug allergy. She had no seizure since she was born.

Family history She is the only child. She was born normally without any complication. No one in her family has experienced a skin disease like her before.

Physical examination Multiple, bead like, brownish papules on the rim of both upper lids with some yellow-white, firm papules, nodules in 2mm diameter(fig1) Her frenulum was infiltrated(fig2)

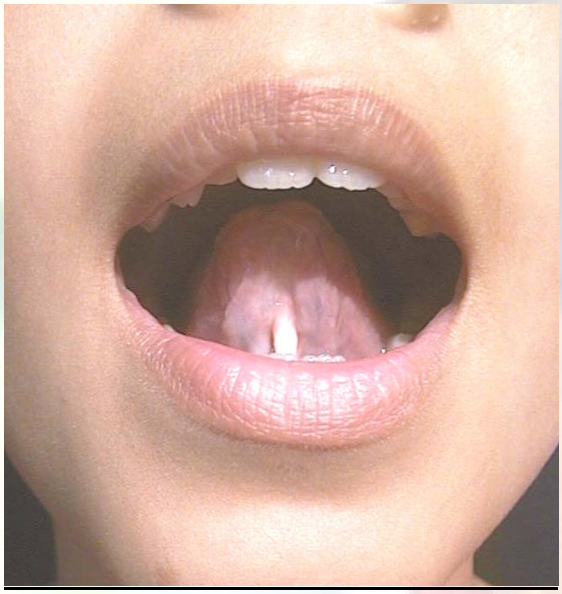

Fig. 7.1

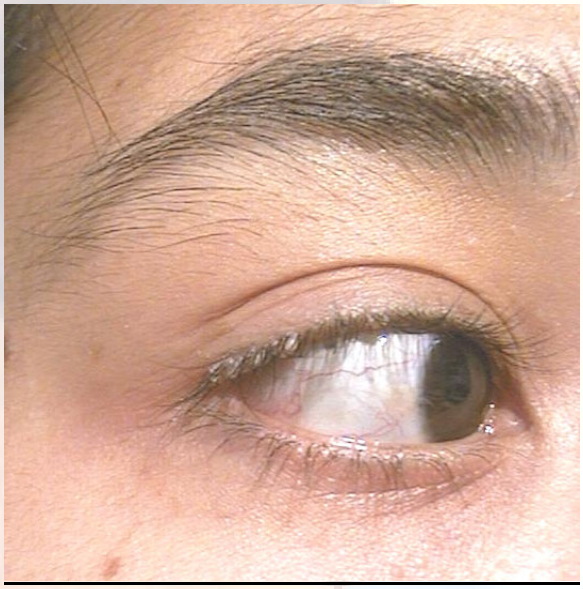

Fig. 7.2

Histopathology Not done.

Diagnosis: Lipoid proteinosis.

Presenter
Consultant

Treatment Advice the patient and her parents about the nature and prognosis of the disease, consult ENT about her vocal hoarseness for evaluation of her pharynx and larynx and also for vocal training.

Discussion Lipoid proteinosis, also known as hyalinosis cutis et mucosae or Urbach Wiethe disease, is a rare autosomal recessive disorder. The most recent study reveals that it link with genome 1q21 disorder and extracellular matrix protein1 gene (ECM1) is found. The disease is characterized by the deposition of an eosinophilic-hyaline like material in the skin, classical feature includes beaded eyelids papules. Larynx, mucous membrane and other internal organs, with a chronic and benign course. However, complications includes laryngeal involvement which may lead to respiratory obstruction and vocal changes. Cerebral involvement may lead to seizure and behavioral changes. Males and females are equally affected (AR) Life expectancy is normal and no effective treatment exists. Anyway, a single case, skin lesions resolved after 3 years of dimethy sulphoxide(DMSO) 60mg/kg/d. But a more recent report showed no benefit in 3 patients with an average treatment duration of 3 years. Etretinate may improve the appearance of the skin lesions.
Multisystemic approach to the lipoid proteinosis should be use (Neurologist, dermatologist, ENT) and the family should be counseled about the risk of having affected offspring.

Reference

1. Hautarzt 1997 Jul;48(7):477-81
2. Wong CK, Lin CS. Remarkable response of lipoid proteinosis to oral dimethyl sulphoxide. Br J Dermatol 1988 Oct;119(4):541-4
3. Joseph J Shaffer. Lipoid proteinosis www.emedicine.com
4. Hamada T, et al. Lipoid proteinosis maps to 1q21 and is caused by mutation in the extracellular matrix protein gene (ECM1). Hum Mol Genet 2002 Apr1;11(7):833-40.