Lipoid proteinosis

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Text books describe Lipoid Proteinosis as a recessively inherited rare disorder, characterised by the infiltration of the dermis and submucosa of the buccal cavity and larynx by a PAS positive hyaline like material; hence the name Hyalinosis Cutis et mucosae. (Urbach and Weithe, 1929).

Patients with this disease are normal at birth. Problems start when they are eight to twelve months old. Their cry becomes hoarse and reduced in volume to a whisper due to infiltrations in the larynx. Recurrent bullae and skin infections occur which heal with scarring.

The skin is yellow, thickened and scarred. Characteristic bead like papules are found at the margin of eye lids. The buccal mucosa, tongue and larynx become infiltrated. Nodules are seen along the vocal cords. The voice becomes hoarse and low volumed with a gravelly character. Para sellar calcifications are seen on skull X-ray in over 50% of cases. Skin biopsy shows the hyaline infiltrations in the dermis. Therefore this is a striking condition which could be diagnosed easily without resorting to sophisticated investigations.

Only about two hundred cases are reported in world literature, all in caucasians of European descent. Presentation of three siblings with this condition in the Southern Province of Sri Lanka by Chularatna and others (Ceylon Medical Journal Vol. 40, No. 1, March 1995) awoke a lot of interest in this Country. Subsequently Jayawardane and others presented five more cases of Lipoid Proteinosis at the annual sessions of the Galle Medical Association 1998. All five of them were form the Southern Province, and three of them were siblings.

Dr. Prasad Kumarasinghe, Dermatologist, Kalutara, has seen three more cases. One of the them was from the Southern Province and the other two from Kalutara District. The two boys from Kalutara District are cousins and are of South Indian Tamil descent, whereas all the other nine cases are Sinhalese.

The apperance of eleven cases of this rare disorder within a few years raises many questions. Is it endemic in this country or was the gene introduced by some of European invaders? What is the exact made of inheritance? Further work is necessary to answer these questions.

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