GENETICS AND GENODERMATOSES

LIPOID PROTEINOSIS: A CASE REPORT IN A FILIPINO MALE.

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Background: Lipoid proteinosis is a rare autosomal recessive disorder due to the loss of function mutation of the ECM1 gene which encodes for extracellular matrix protein 1 (ECM1), a secretory protein which promotes angiogenesis and is a negative regulator of endochondral bone formation. Also known as Urbach–Wiethe disease or hyalinosis cutis et mucosae, this genodermatosis presents with bead-like papules on the eyelid margins (monoliform blepharosis), acneiform scarring, waxy papules, hyperkeratotic or verrucous plaques and hoarseness of voice. Deposition of hyaline material on the skin and internal organs can be life-threatening. Since its discovery in 1929, there have been approximately 300 cases reported worldwide.

Observation: We present a case of a 23-year-old Filipino male presented with a long history of skin fragility, chronic recurrent verrucous and waxy plaques on the face and body, accompanied by monoliform blepharosis, diffuse alopecia of the scalp and hoarseness of voice. 4-mm skin punch biopsy was done and revealed hyaline material on the papillary dermis, surrounding blood vessels and adnexal structures. There is a mild superficial and mid-dermal perivascular inflammatory infiltrate of lymphocytes. Special stain such as Alcian blue revealed hyaline deposition. Reduction of the waxy papules and verrucous plaques was done using CO2 laser ablation with the following parameters: ultra-pulse mode, fluence 40 J/cm2, pulse duration 20 J/cm2 which resulted in partial resolution of some lesions on the nape and arms.

Key Message: Early recognition of the signs and symptoms is essential to the clinician and underscores the importance of multidisciplnary approach in the management of the symptoms and course of Lipoid proteinosis.