GENETICS AND GENODERMATOSES

A RARE CASE REPORT OF LIPOID PROTEINOSIS OF URBACH WIETHE , WITH EYE CATCHING UNIQUE FEATURES

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Background: Lipoid proteinosis also known as Urbach – Weithe disease and Hyalinosis cutis et mucosae is a rare, autosomal, recessively inherited disorder, characterized by hoarseness from early infancy, together with various cutaneous manifestations, such as, acneform scarring, waxy papules, eyelid beading (moniliform blepharosis). As well as, non-cutaneous manifestation attributed to infiltration of hyaline-like material in the skin, larynx, and various organs. The hyaline-like material is Periodic-Acid Schiff (PAS) positive and diastase resistant and is believed to be the result of the deposition of non-collagenous proteins and glycoprotein. Although this rare autosomal recessive disorder has been described in the literature, its occurrence is rare in India. We herewith report this case for its rarity and very typical diagnostic features.

Observation: A 16-year-old girl born to second-degree consanguineous parents presented with multiple asymptomatic variable raised skin-lesions and hoarseness of voice since early-childhood. She had no history of seizures, visual disturbances, photosensitivity, or respiratory obstruction, but presented with restricted tongue movement and speech impairment. On examination, the patient had hoarseness of voice and numerous skin-colored waxy papules on the face, forehead. Pearly beaded papules were seen at both the upper and lower eyelid margins (so-called moniliform blepharosis). Hyperkeratotic wart-like lesions were seen on the elbows and knees. Enlarged tongue with irregular pearly white infiltration. Rhinolaryngoscopy revealed diffuse Nodular deposition involving Arytenoids, epiglottis and anterior two thirds of Vocal cords responsible for the hoarseness in her voice. Histopathology - Acellular PAS positive diastase resistant eosinophilic hyaline like material is seen deposited in the dermis surrounding the blood vessels and appendages.

Key message: The characteristic beaded-papules on eyelid -margin, acneform-scarring and hoarseness supported by histology pointed to the rare diagnosis of Lipoid-proteinosis.