GENETICS AND GENODERMATOSES

SCHIMMELPENNING-FEUERSTEIN- MIMS SYNDROME A RARE ENTITY ASSOCIATED WITH RARER ASSOCIATIONS

Priyanka Ghuge [(1)] - Uday Khopkar [(1)]

Seth G. S. Medical College And K. E. M. Hospital, Dermatology And Venereology, Mumbai, India [(1)]

Background: Schimmelpenning-Feuerstein-Mims syndrome, a type of epidermal nevus syndrome, characterised by organoid nevi, abnormalities of the central nervous system, eyes, oral lesions and skeletal defects.
Observation: A 14 year old female presented with dark thick lesions on the face and neck, bald patch on the scalp, deformed teeth and lesions in the mouth and eyes, since birth.

Examination: 2 well defined hyperpigmented verrucous hyperkeratotic plaques over the left upper face extending into the scalp and the neck. Well defined smooth non scarring 3 cm patch of alopecia on the frontal scalp. Erythematous infiltrating plaques in conjunctiva, malformed deciduous teeth, lobulated gum hypertrophy and soft lobulated plaques on the tongue with no tendency to bleed on minimal handling.
General examination: global developmental delay, facial asymmetry, left sided torticollis and limb length discrepancy.
Histopathology verrucous plaque: orthohyperkeratosis, acanthosis, massive papillomatosis
Histopathology alopecia patch: absent hair follicles with orphan arrector pili muscle.
Histopathology tongue: acanthosis and papillomatosis suggestive of mucosal counterpart of the epidermal nevus.
Ophthalmological evaluation: bilateral limbal lipodermoids, vascularised corneal opacities in right eye.
MRI brain: hemimegalencephaly right occipital lobe, sphenoid wing dysplasia with arachnoid cysts in right anterior temporal lobe
Fibroblast growth factor 23: highly elevated: 228.1. No evidence of hypophosphatemia or history of bone fractures.
Patient was partially operated for the nevus. Fgf23 levels repeated after 6 weeks were decreased.

Key message: We present this case for the presence of concurrent presence of mucosal epidermal nevus with cutaneous lesions, limbal dermoids which is defined but a very rare association and raised Fibroblast growth factor 23 which showed a significant reduction following excision of a part of the nevus supporting the theory that the nevus tissue/ skin to

be the source ultimately leading to Cutaneous skeletal hypophosphatemia syndrome.A multidisciplinary approach and awareness is important for timely diagnosis and treatment.