

4th International Conference on

WOUND CARE, WOUND NURSING TISSUE REPAIR & REGENERATIVE MEDICINE

October 05-06, 2018 | Los Angeles, USA

Epidermolytic hyperkeratosis with hypogonadism and growth retardation

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Introduction: Epidermolytic ichthyosis (EI), formerly known as epidermolytic hyperkeratosis or bullous congenital ichthyosiform erythroderma is a rare genodermatosis inherited most commonly in an autosomal dominant fashion but may occur as a spontaneous mutation. It is classified under the non-syndromic ichthyosis.

Case: A 30 year old adult male patient presented with generalized involvement of the body with erythematous dry, hyperkeratotic plaques associated with diffuse scaling and short stature. It was associated with diffuse palmoplantar keratoderma. There were history of bullous lesions in childhood. He was born out of non-consanguineous marriage through normal vaginal delivery. Family and past history was non significant. Testicular size were below the normal average and secondary sexual characters were absent.

Investigations: Skin biopsy, hormonal profile, physical measurements were taken.

Diagnosis: Epidermolytic Hyperkeratosis [PS(palms/soles involved)-2 type] with secondary hypogonadism and dwarfism.

Conclusion: Although epidermolytic hyperkeratosis is not associated with any underlying systemic abnormality, there are case reports with associated hypogonadism and may require early paediatric endocrinologist consultation for a better quality of life.

Biography

Kaur A has completed her MBBS at the age of 24 years from Government Medical College and Hospital, Chandigarh (UT), India. She is currently pursuing her post graduation junior residency i.e. MD dermatology, venereology, and leprosy from Government Medical College And Hospital, Chandigarh (UT), India. She has 2 case reports that are under publications. She has presented around 5 posters and 2 oral presentations in different national conferences.

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