GENETICS AND GENODERMATOSES

ANNULAR EPIDERMOLYTIC ICHTHYOSIS: A RARE PHENOTYPIC VARIANT OF BULLOUS CONGENITAL ICHTHYOSIFORM ERYTHRODERMA

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Background: Bullous congenital ichthyosiform erythroderma/EHK is an autosomal dominant disorder characterized by blistering, erythroderma and hyperkeratosis starting at birth and shows epidermolysis on microscopy. Annular epidermolytic hyperkeratosis is a rare phenotypic variant of EHK, characterised by intermittent bouts of annular and polycyclic, erythematous, scaly plaques on the trunk and proximal extremities.

Observation: A 26-year-old female, born to non-consanguineous parents, presented with classical history of bullous lesions, erythroderma and hyperkeratosis starting since birth. But since last 3 years, she also started developing erythematous to hyperpigmented round to oval patches with scales over trunk, thighs and inframammary areas. Many showed an annular configuration. Such lesions appeared in an episodic manner every 3-4 weeks. She has one affected male child with EHK. There was no history of colloidon membrane at birth in the mother and child. Histopathology from the annular plaque and ridged hyperkeratotic plaques revealed hyperkeratosis, acanthosis, thickened granular layer and cytoplasmic vacuolization in upper spinous and granular layers. She was started on ciclosporin 3.5mg/kg body weight with initial improvement but had a recurrence after 1 month. She was then started on acitretin 0.5 mg/kg/day after detailed counselling regarding family planning and there was remarkable improvement 3 weeks later with no recurrence till now.

Key message: The distinct phenotype seen in our patient has only been described in eight families thus far in the literature. It was first reported by Sahn et al. in 1992 who coined the term annular epidermolytic ichthyosis. Similar cases were reported by Suga et al. in 1998 and Sybert et al in 1999. Molecular analysis of these families revealed mutations in K10 and K1 respectively. We report this case as it is a rare morphologic presentation of bullous congenital ichthyosiform erythroderma and probably the first from India.