

ICHTHYOSIS HYSTRIX – A RARE CASE REPORT

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Article Info <i>Received 15/04/2016</i> <i>Revised 27/05/2016</i> <i>Accepted 2/06/2016</i> Key words: Ichthyosis hystrix, CM type, Keratin1 mutation.	ABSTRACT Ichthyosis hystrix is an uncommon, heterogenous group of dermatoses which differs both genetically and clinically. It may be inherited as autosomal dominant trait or occur sporadically. It is characterised by spiny hyperkeratotic scales which can vary in expression from a generalised involvement to localised and nevoid forms. We, here by report a case of ichthyosis hystrix in a 9 year old girl, which is sporadic in onset involving face, trunk and extremities.
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INTRODUCTION

Ichthyosis hystrix, which encompasses multiple rare heterogenous disorders in ichthyosis family, presents with massive hyperkeratosis and spiny scales commonly over trunk and extensor aspects of extremities. It can also be used to indicate localised and linear warty epidermal nevi. The word ichthyos means fish like scales and hystrix means resemblance similar to spines of porcupine: hence termed as porcupine disease. Five different clinical phenotypes have been described, namely Brocq, Rheydt, Bafverstedt, Lambert, and Curth-Macklin (CM) type.

CASE REPORT:

A 9 year old girl presented to our department with complaints of multiple dark raised, persistent scaly skin lesions over face, neck, upper and lower limbs since 2 years of age, which was initially localised, gradually progressed to involve the entire body. The child was born to non consanguineous parents by full term normal vaginal delivery. No history collodion membrane formation or erythema or blistering at birth. Growth and developmental milestones were appropriate for age, with an exception of difficulty in learning. No history of symptoms or signs suggestive of systemic involvement and no history of similar complaints in any of the family members. On dermatological examination, bilaterally symmetrical thick

spiny, hyperkeratotic papules which coalesce to form linear pigmented cobblestone-like scaly plaques were present over face, neck, upper and lower limbs and trunk. Scaling was seen more over the flexures. The lesions followed the lines of Blascko. Palms, soles, nails and genitalia were spared. No abnormalities were detected in ENT, eye, neurological or skeletal system. Skin biopsy of a lesion over the back revealed – hyperkeratosis, moderate acanthosis with elongated rete ridge and regular alteration of slightly raised parakeratotic areas without a granular layer and, slightly depressed orthokeratotic areas with prominent granular layers. Inflammatory infiltrates were noted in the dermis. Hence we concluded to the diagnosis of Ichthyosis hystrix.

DISCUSSION:

Ichthyoses involve a diverse group of keratinisation disorders, resulting from abnormal epidermal differentiation or metabolism. In general, they constitute extensive scaling and areas of skin thickening (Vijayeeta Jairath *et al.*, 2015) Ichthyosis hystrix (IH) can be inherited as an autosomal dominant trait or occur sporadically (Penrose LS and Stern C, 1957). It may present at birth or may have a delayed onset in infancy or early childhood. It is considered as a variant of epidermal

nevi with widespread bilateral distribution (Surajit Nayak *et al.*, 2013). It is also referred to as systematised verrucous epidermal nevus or Porcupine man since it is characterised by the presence of spiny hyperkeratotic scales, simulating bullous ichthyosiform erythroderma, but differs in the absence of blistering, mild or absent erythroderma and specific ultrastructural and histological features.

IH and epidermolytic hyperkeratosis are ichthyosis involving pathology in keratinisation of skin. They occur as a result of mutations of keratin 1 (K1) and keratin 10 (K10) genes which lead to an impairment of keratin intermediate filament (KIFs) network assembly in the cytoplasm of keratinocytes (Fonseca *et al.*, 2013). The differentiating keratinocytes exhibit ultrastructural changes such as presence of peripheral shells, perinuclear vacuolization and formation of binucleated cells.

The variations in phenotypic expression, arising from a single gene mutation are supposed to be resulting either from the involvement of different parts and specific protein domains of the gene or a combination of unidentified genetic modifying factors and environmental influences. IH Brocq type and EHK are due to mutations in critical position of rod domain in K1 and K10 genes, respectively. K1 mutations cause PS phenotype, K10 mutations give rise to NPS phenotype and Connexin 26 mutation leads to IH Rheydt type. IH-CM and Lambert type occur due to mutations in tail domain of K1 and K10 genes, respectively (Wang *et al.*, 2007).

Clinically, IH presents as verrucous or porcupine-like hyperkeratotic scales over extensor aspect of extremities and trunk with lesser extent towards flexures. It can also be associated with scaling over scalp and palmoplantar keratoderma. Nevoid lesions may also follow Blaschko's lines (Nayak *et al.*, 2013). Due to significant phenotypic heterogeneity, IH is subdivided into 5 types based on the pattern of distribution namely, Brocq, Rheydt, Bafverstedt, Lambert, and CM types. In Brocq type, erythroderma and blistering may precede the development of hystrix scales. Rheydt type shows hyperkeratosis of face and extremities with deafness, which is now referred as HID syndrome (Hystrix, Ichthyosis and Deafness). Bafverstedt type was documented in a single case with follicular hyperkeratosis and mild involvement of palms. Lambert type illustrates the first recorded cases in 1730s; the members affected were being referred to as "porcupine men". It presents as generalised hystrix scaling which spares palms and soles (NPS). CM type shows features similar to Lambert type, with involvement of palms and soles (PS).

IH may be associated with a variety of cutaneous, ocular, skeletal, neurological, cardiovascular and urogenital developmental anomalies, which is termed as epidermal nevus syndrome (ENS) (Solomon LM, Esterly NB, 1975). Under light microscopy, hyperkeratosis, patchy parakeratosis, acanthosis and papillomatosis are seen. The upper dermis shows vacuolated and binucleate keratinocytes. Granular layer shows reduced thickness and contains rounded and few vacuolated cells. IH should be differentiated from bullous type of epidermolytic hyperkeratosis, which presents at birth with blistering and eventually develops hystrix spines predominantly over flexural creases. EHK depicts characteristic, vacuolar degeneration of upper epidermis with large keratohyaline granules and perinuclear halo.

This case report represents Curth-Macklin (CM) type of IH with Blaschkoid pattern but sparing palms and soles, which is relatively rare. So far, only two large families and a few sporadic cases of IH-CM have been documented, since its first description in 1954 (Yusuf *et al.*, 2009). Despite the fact that previous case reports illustrate the predominance of hyperkeratosis in extensor surfaces of limbs, our patient showed flexural involvement, which had been earlier reported in only few cases (Biswas *et al.*, 2014). Although, flexural involvement is more distinctive of EHK, it is ruled out by reason of no history of blister formation and lack of epidermal vacuolar degeneration in skin biopsy. Even though the child had extensive cutaneous disfigurement, she showed no systemic involvement and has had normal growth. Treatment of IH is confined to topical and systemic retinoids (Basler *et al.*, 1978; Richardson *et al.*, 2006) either alone or in combination with keratolytics (salicylic acid, urea-containing compounds or propylene glycol) to reduce hystrix scaling and keratoderma. Topical tacrolimus may also be beneficial. Despite the chronicity and protracted course of disease, scaling may improve or stabilise as the age progresses.

CONCLUSION

This case is reported because of its rare occurrence and generalised involvement with Blaschkoid pattern.

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CONFLICT OF INTEREST

The authors declare that they have no conflict of interest.

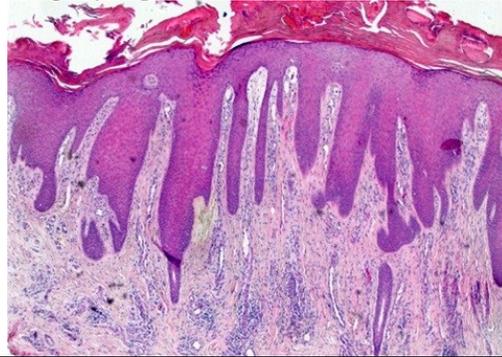
Figure 1: Clinical photograph showing verrucous pigmented plaques present along the lines of Blaschko in bilateral upper limbs and trunk.



Figure 2: Clinical photograph showing verrucous plaques over the back.



Figure 3: Histopathology showing hyperkeratosis with elongated rete ridges and slightly raised parakeratotic areas without a granular layer and slightly depressed orthokeratotic areas with prominent granular layer.



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