

# A sporadic case of ichthyosis hystrix: A rare entity

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**Abstract** The term ichthyosis hystrix encompasses several rare ichthyosiform disorders characterized by massive hyperkeratosis with an appearance like spiny scales. This is a rare form of ichthyosis with autosomal dominant inheritance and a very few cases have been reported. We report a case of ichthyosis hystrix in a 22-year-old male patient with hyperkeratotic, dry, warty, scaly excrescences on most of the body surface in a linear pattern. Palms, soles and nails were spared and family history was negative. The case is being reported in view of the rarity of disease and its sporadic occurrence.

**Key words**

Ichthyosis hystrix, hyperkeratosis, sporadic

## Introduction

Ichthyosis hystrix is a group of rare conditions, characterized by spiny, hyperkeratotic scales in which blistering does not occur, erythroderma is mild or absent and localized and nevoid forms are more common.<sup>1</sup> The term is derived from the Greek roots for fish (*ichthys*) and porcupine (*hustriks*). It is inherited either as autosomal dominant or as sporadic variety.<sup>2</sup>

The lesions mostly occur on the extensor aspects of arms and legs. Patients with extensive involvement resemble those with epidermolytic hyperkeratosis with or without palmar/ plantar involvement, with verrucous or porcupine-like hyperkeratosis.<sup>3</sup>

The term has also been employed to describe localized and linear, warty epidermal nevi sometimes associated with mental retardation, seizures or skeletal anomalies. Alopecia and hair and nail abnormalities, as well as, inner ear

deafness are also seen in these patients.

## Case Report

A 22-year old male patient presented in our outpatient department with multiple, nonpruritic, warty lesions on most of the body surface for 20 years. The father reported that the patient was born out of non-consanguineous marriage, full term by a normal vaginal delivery. The lesions were first noted at birth as a hyperpigmented, papular lesions on the medial aspect of right thigh, they gradually spread to the whole body and became warty and scaly. There was no history of blistering. The patient had no history of seizures, mental retardation, hearing or visual abnormalities. All developmental milestones were normal. None of the family members had any similar complaints.

General physical and systemic examination was normal.

On cutaneous examination, multiple, linear, hyperkeratotic, verrucous, dry, hyperpigmented papules and plaques were seen over the angle of

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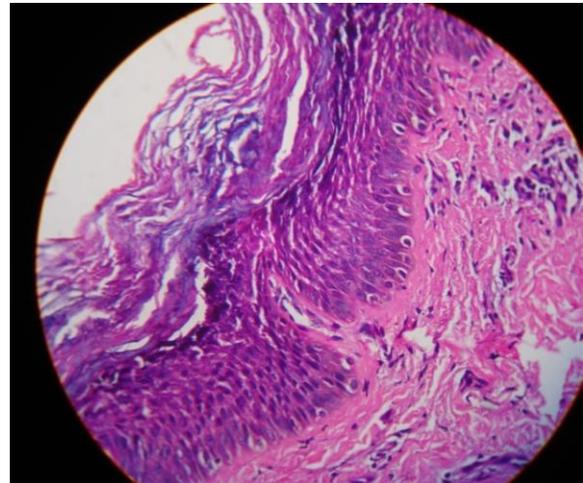
**Figure 1** Diffuse, hyperkeratotic, warty, dry papules and plaques.



**Figure 2** Lesions in a linear pattern, along the lines of Blaschko.



**Figure 3** Similar lesions on the posterior trunk with areas of normal skin.



**Figure 4** Histopathological examination showed epidermal hyperplasia with papillomatosis and epidermal digitations, thickened granular layer with coarse keratohyalin granules and marked vacuolization of the granular and upper spinous layers, thickened stratum corneum.

mandible, ears, neck, anterior and posterior trunk and all extremities including the dorsal surfaces along the lines of Blaschko (**Figure 1** and **2**). There were a few areas of normal skin noted on the right side of the trunk and extensor aspect of lower limbs (**Figure 3**). Scalp, palms and soles were spared, so was most part of the face. Nails were normal. No dental

abnormalities were seen. Mucosal surfaces showed no abnormality.

Routine laboratory examination including complete blood count, urinalysis, liver function tests, renal function tests and chest X-ray showed no abnormal findings.

Histopathological examination of the skin lesion showed a focus of epidermal hyperplasia with papillomatosis and epidermal digitations. The granular layer was thickened and showed coarse keratohyalin granules with marked vacuolization of the granular and upper spinous layers. The stratum corneum was markedly thickened and lamellated. These findings were consistent with ichthyosis hystrix (**Figure 4**).

On the basis of the above constellation of symptoms (history, morphology of lesions, histopathological findings), a diagnosis of ichthyosis hystrix was made. The patient was managed with oral isotretinoin, topical keratolytics (urea and lactic acid) and emollients.

## Discussion

Ichthyosis hystrix is a descriptive name for a heterogeneous group of keratinization disorders sharing similar clinical features of massive, spiky or verrucous lesions clinically and epidermolytic hyperkeratosis histologically.<sup>4</sup> It was first described between 1731-1851 in the Lambert family of Suffolk affecting 11 family members in four generations, the first one being Edward Lambert (along with his descendants, known as the porcupine men).

In 1902, Brocq<sup>5</sup> described ichthyosis hystrix as an atypical form of congenital bullous ichthyosiform erythroderma. It was later described by various names by other authors like nevus verrucous, systematized epidermal nevus and epidermolytic hyperkeratosis. Gianotti,<sup>6</sup> in 1980, classified it as a localized form of

**Table 1** Types of ichthyosis hystrix [7].

Type	Characteristic features
1. Curth and Macklin	Prominent extensor involvement, severe palmoplantar keratoderma,
2. Lambert	Childhood onset, palms and soles spared
3. Rheydt	Generalized warty hyperkeratosis with deafness
4. Bafverstedt	Striking scaling over face

congenital epidermolytic hyperkeratosis. Curth and Macklin described two teenage brothers with similar presentation of ichthyosis. Affected relatives spanning five generations had a variable phenotype and the condition was regarded as autosomal dominant.

The types of ichthyosis hystrix include Brocq, Lambert, Curth-Macklin, Rheydt and Bafverstedt<sup>7</sup> (**Table 1**). A study shows mutation in variable tail (V2) domain of keratin 1.<sup>8</sup> We could not investigate for the same due to lack of this facility at our centre.

The Curth and Macklin (IHCM-Ichthyosis Hystrix of Curth and Macklin) subtype is characterized by severe palmoplantar keratoderma occurring in a striated or striped pattern. Spine-like, brown lesions are seen over the whole body, being more prominent on the extensors. Histologically, binucleate keratinocytes are seen. In our patient, the palms and soles were spared.

In Lambert type, onset is delayed till childhood. Phenotype and histology is similar to IHCM but palms and soles are spared.

In Rheydt type (also known as HID-hystrix like ichthyosis and deafness), generalized warty hyperkeratosis and deafness are seen. Connexin 26 mutation has been noted.<sup>9</sup> Our patient had no deafness.

In Bafverstedt, striking hystrix scaling of face is seen. Our patient had very few lesions on the facial skin.

Histopathological examination shows severe orthohyperkeratosis, patchy parakeratosis, acanthosis, and papillomatosis with scattered vacuolated and binucleate keratinocytes in the upper epidermis. In our patient, histopathology revealed hyperkeratosis, acanthosis, marked vacuolization of granular and upper spinous layers with coarse keratohyalin granules.

We prescribed our patient with topical keratolytics, emollients and systemic retinoids. The treatment showed some beneficial result.

We decided to report this case because of the rarity of disease, especially sporadic occurrence. Less than five cases have been reported from India. To the best of our knowledge, this is the first case of Ichthyosis hystrix being reported from Jharkhand, India.

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