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PORCUPINE LIKE ICHTHYOSIS HYSTRIX: A CASE REPORT

Dermatology

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Introduction:
Ichthyosis hystrix was described first in 1731 as an uncommon case of distempered skin by John Machin[1,2]. The same man with a son having the same condition was described by the name Porcupine Man in the 1755.The name hystrix is derived from the Greek word for porcupine.
It is a term used for keratinization disorders having spiky, massive verrucous hyperkeratosis and histologically showing epidermolytic hyperkeratosis[3]. It is a rare variant of ichthyosis and may be associated with neurological deficit and deafness. It is considered to be an autosomal dominant disorder with some de novo cases reported[4].

Case report:
An 11 year old male patient born of a non-consanguineous marriage presented with complaints of generalized severely thickened skin since 3 months of age. The patient was normal at birth and started developing hyperpigmented papular lesions which first started over the upper limbs and then progressed to involve the lower limbs, trunk, face and the scalp. This was followed by development of porcupine like spikes over the skin. Developmental history was normal. No history of similar complaints in the family members.

On examination hyperkeratotic skin with porcupine like scales arranged in rows were present over the face, scalp, bilateral upper limbs, gluteal area and lower limbs, chest and upper back (Figure 1). The scales were arranged in a mosaic pattern following the lines of Blashko. Alopecia was present over the affected areas of the scalp (Figure 2). Hyperpigmented papular lesions were present over the abdomen,malar area and the back. Areas of spared skin were present over the trunk and the lower limbs. Foul odour was present. Hyper linearity of palms was seen. Hyperkeratotic plaques were seen over the soles. Few hyperpigmented papules were seen over the penis. Oral cavity was normal. There was no hearing loss nor were there any skeletal or ophthalmological abnormalities. Laboratory parameters were within normal limits. A wet mount for fungus from scales taken from the scalp was negative.

Histopathological examination of a biopsy taken from the porcupine like scaly area revealed only massive hyperkeratosis (Figure 3a). Hence a biopsy was taken from the pigmented papular lesion and it showed hyperkeratosis, vacuolated granular layer, increased melanocytes along the basal layer of the epidermis (Figure 3b). The papillary dermis showed sparse lymphocytic infiltrate. All these changes are suggestive of epidermolytic hyperkeratosis

Based on the history, clinical features and histopathology, a diagnosis of ichthyosis hystrix was made.

Discussion:
Ichthyosis hystrix is of various types namely Lambert, Curth and Macklin, Brocq, Bäfverstedtand Rheydt[5]. This patient on the basis of the clinical features and histology has Curth and Macklin type. These patients have extensive massive spiny hyperkeratotic plaques covering the whole body and have palmoplantar involvement. This is most often due to pathogenic mutations in KRT1 affecting the variable tail domain (v2) of keratin 1[6].

The extensive scaling leads to secondary bacterial infection. The closure of the scales leads to anaerobic infection leading to a foul odour.

Since no relatives were previously affected in this case, it can be considered to be due to a mutation. The scales being arranged in a mosaic pattern along Blaschko's lines is suggestive of mosaicism. This case is more suggestive of type 1 mosaicism in which one allele of the gene which is involved is mutated leading to the disease to present in a mosaic form.

Treatment needs to be started at an early age for effective results. The prognosis is poor.Systemic retinoids especially acitretin needs to be given for a long duration. Surgical management can be done for a few lesions but is difficult in case of extensive disease. Regular soaks and baths need to be given for maintaining hygiene.

This patient was started on systemic acitretin 10 mg twice a day with considerable improvement (Figures 4&5), topical retinoic acid 0.05% for the pigmented papular lesions. Systemic antibiotics were also given for the secondary bacterial infection. The patient was also given potassium permanganate soaks.

This case has been reported for the childhood presentation of severe disease and sporadic occurrence. To our best knowledge this is the fourth case if ichthyosis hystrix reported from India and the first to have such severe disease with extensive porcupine like scaling in childhood.

Conflict of interest: None

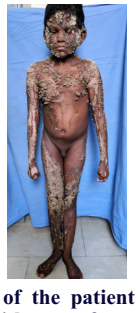

Figure 1: Frontal view of the patient showing massive spiky hyperkeratotic plaques with rows of porcupine like scales over the face, chest, and bilateral upper and lower limbs.

Figure 2: Lateral view of the patient showing a closer view of the porcupine like scaling over the face and hair loss over the affected region of the scalp.

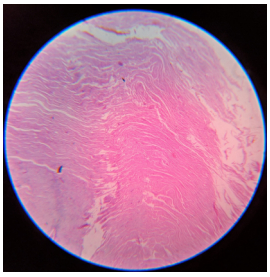

Figure 3a: 10x view of the formalin fixed H&E stained section of the skin taken from the spiky plaque showing massive hyperkeratosis.

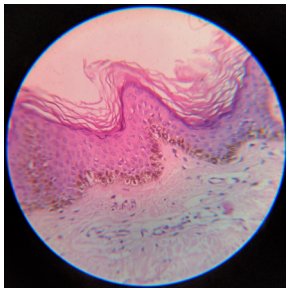

Figure 3b: 40x view of the formalin fixed H&E stained section of the skin taken from the pigmented papule over the chest showing hyperkeratosis, vacuolated granular layer, acanthosis and increased melanocytes along the basal layer of the epidermis. The dermis showed sparse lymphocytic infiltrate.

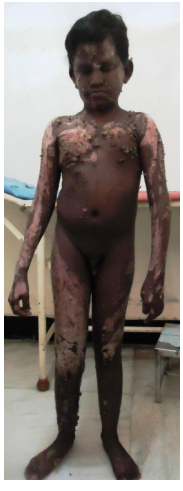

Figure 4: Frontal view of the patient post-treatment showing considerable reduction of the scaling.

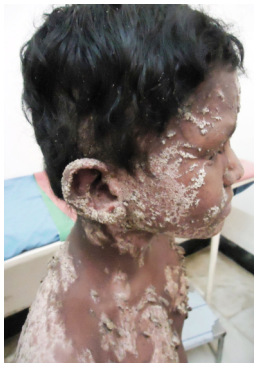

Figure 5: Lateral view of the patient post-treatment showing reduction of the scaling. The foul odour had also come down.

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