VOLUME-7, ISSUE-8, AUGUST-2018 • PRINT ISSN No 2277 - 8160

Original Research Paper

Dermatology

ICHTHYOSIS HYSTRIX: A CASE SERIES.

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ABSTRACT Ichthyosis Hystrix is a keratinization disorder which is usually inherited in an autosomal dominant manner but sporadic cases may also be seen. It is an uncommon disorder and may have varied clinical presentations. It usually is characterized by hyperkeratotic scales which can either be generalized or even localised and naevoid forms are also see. We report 3 cases of Ichthyosis Hystrix having different age groups of presentation and different distribution.

KEYWORDS : Ichthyosis Hystrix, keratinization disorder, hyperkeratotic scales.

Introduction:

Ichthyosis Hystrix was initially described by John Machin as an uncommon case of distempered skin [1]. Ichthyosis Hystrix is a term used for a group of keratinization disorders presenting with massive verrucous hyperkeratosis clinically and epidermolytic hyperkeratosis histologically[2] When generalized it is also referred to as systematized epidermal naevus[3] and it usually has an autosomal dominant mode of inheritance though a few sporadic cases have also been reported[4].

Case reports:

Case 1:

The first patient was a 11-year-old male who presented with severe generalized severely hyperkeratotic skin since 3 months of age. The patient was apparently normal at birth and subsequently developed hyperpigmented papular lesions which initially started over the upper extremities and later spread to involve the trunk, face, scalp and lower limbs which was then followed by developed of porcupine like scales. There were no developmental abnormalities.

On clinical examination hyperkeratotic skin with porcupine like scales following Blaschko's lines were seen over all the above areas (Figure 1). Moreover, hyperpigmented papules were seen over the malar area, abdomen and back. Hyperkeratotic plaques were present over the soles and alopecia was seen over the affected areas of the scalp (Figure 2). A severe foul odour was present.

Histological examination of the porcupine like scales showed massive hyperkeratosis. Biopsy taken from the pigmented papular lesion showed hyperkeratosis, increased melanocytes in the stratum basale and vacuolated granular layer (Figure 3). Papillary dermis revealed a sparse lymphocytic infiltrate.

Case 2:

The second patient was a 9-year-old female who presented with complaints of pigmented scaly lesions over the face, neck, upper and lower extremities since the age of 2. It was initially localized and later spread to involve the entire body and was persistent. Growth and development were normal for age with a mild difficulty in learning which could also be due to the skin condition.

On clinical examination bilaterally symmetrical spiny, hyperkeratotic papules coalescing to form pigmented scaly plaques were seen over the face and neck, trunk, upper and lower extremities following Blaschko's lines (Figure 4). Flexural scaling was present with sparing of the palms, soles, genitalia and nails.

Histopathological examination (Figure 5) revealed hyperkeratosis, papillomatosis, raised parakeratotic areas with agranulosis and depressed orthokeratotic areas with a prominent granular layer.

Case 3:

The third patient was a 35-year-old male who presented with complaints of pigmented raised lesions all over the body since the last seven years. Initially a small papule developed over the right gluteal area at 2 months of age. Subsequently there was spread and enlargement of lesions over the entire body since the last 7 years. Pruritus was present. History of seizures was present.

On clinical examination symmetrical pigmented adherent scaly plaques in a blashkoid distribution were seen in a generalized pattern with involvement of both the flexor and the extensor areas and the scalp (Figure 6). Severe verrucosity was seen over the lower extremity lesions (Figure 7). There was no involvement of the genitalia, nails, palms and soles. Histopathological examination revealed hyperkeratosis, acanthosis and papillomatosis (Figure 8)

Family history was negative in all 3 cases. Systemic examination was normal in all these cases. The biopsy changes in all cases were suggestive of epidermolytic hyperkeratosis. Based on the clinical and histological features, a diagnosis of Ichthyosis Hystrix was made in all 3 cases. The first case was treated with oral acitretin 10mg per day with considerable improvement.

Discussion:

Ichthyosis Hystrix is an uncommon genodermatosis and occurs due to a defect in the keratin 1 coding gene[5]. The term Hystrix is derived from a Greek word meaning 'porcupine-like' due to the presence of spine like hyperkeratotic verrucous scales.

There are five types of Ichthyosis Hystrix namely Lambert, Brocq, Curth-Macklin, Rheydt and Brocq and Bäfverstedt[7].

Erythroderma and blistering prior to the onset of lesions is present in the Brocq type. Deafness is seen in the Rheydt type. Diffuse facial involvement along with follicular hyperkeratosis is seen in Bafverstedt type. The genitalia, palms, soles and face are spared in the Lambert type.

Diffuse lesions all over the body along with involvement of palms and soles is seen in the Curth-Macklin type. These three patients have the Curth and Macklin type based on the histological and clinical features. Relatives were not affected in all the cases and hence all of can be considered to be due to a mutation. Since the scales were arranged along the Blaschko's lines in a mosaic pattern, it is suggestive of mosaicism. A foul odour may occur in these patients due to closure of scales leading to anaerobic infection.

Treatment at an early age is necessary and the condition has a poor prognosis. Long term systemic retinoid therapy (acitretin) is required.

Conclusion:

All the 3 cases reported in this case series were of the Curth-Macklin type of Ichthyosis Hystrix but presenting in different age groups with different presentations. Ichthyosis Hystrix may have different presentations deviating from the classic presentation even in the same type of the condition.

Figure 1: Frontal view of case 1showing massive spiky hyperkeratotic plaques along with porcupine like scales in rows.

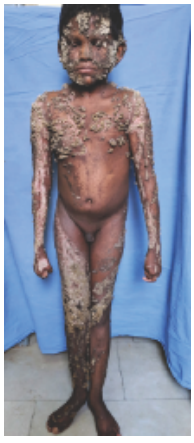


Figure 2: Lateral view of the patient in case 1 showing alopecia over the scalp in the affected areas.

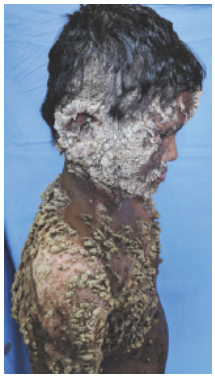


Figure 3: Histopathological examination of the pigmented papule over the chest in case 1 showing hyperkeratosis, vacuolated granular layer, acanthosis and increased melanocytes along the basal layer of the epidermis. The dermis showed sparse lymphocytic infiltrate.

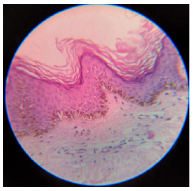


Figure 4: Clinical photograph of case 2 showing verrucous plaques over the back arranged in a blaschkoid pattern.

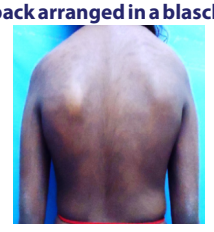


Figure 5: Histopathological picture of case 2 showing hyperkeratosis with papillomatosis and slightly raised parakeratotic areas with agranulosis and slightly depressed orthokeratotic areas with a prominent granular layer.

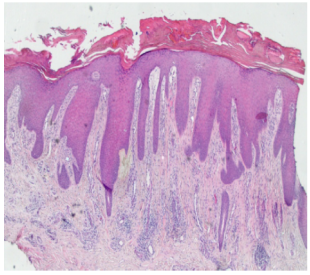


Figure 6: Clinical picture of case 3 showing symmetrical, pigmented, verrucous plaques over the upper extremities and the trunk arranged in a blaschkoid pattern.

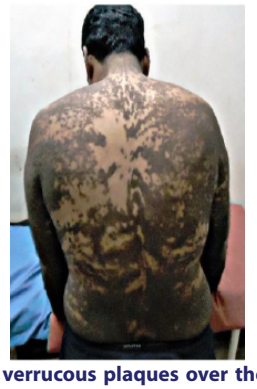


Figure 7: Severely verrucous plaques over the bilateral lower extremities seen in case 3.

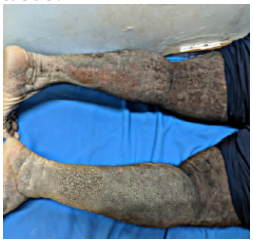


Figure 8: Histopathological examination of case 3 revealing hyperkeratosis, acanthosis and papillomatosis.

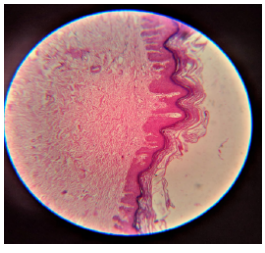
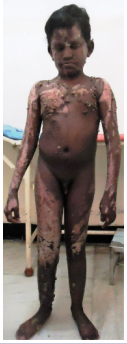


Figure 9: Frontal view of the patient in case 1 showing significant improvement of the lesions on treatment with acitretin.



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