

## Non-bullous Ichthyosiform Erythroderma with Mosaic Turner's Syndrome - a Rare Association

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### Abstract

The ichthyoses are a heterogeneous group of skin diseases characterized by generalised scaling. Non-bullous Ichthyosiform Erythroderma (NBIE) or congenital ichthyosiform erythroderma is a rare and usually severe autosomal recessive inflammatory ichthyosis. Turner's syndrome is a condition caused by numeric and structural abnormalities of the X chromosome. In individuals with mosaic Turner's syndrome, the second X chromosome is not entirely missing. In addition to the expanding list of syndromes that combine with ichthyosis. We report non-bullous ichthyosiform erythroderma in a patient with mosaic Turner's syndrome. To our knowledge no such association has been reported in literature to date.

**Keywords:** Non-bullous ichthyosiform erythroderma; Turner's syndrome; Mosaicism

### Introduction

The ichthyoses are a heterogeneous group of skin diseases characterized by generalised scaling [1]. Non-bullous ichthyosiform erythroderma (NBIE) or congenital ichthyosiform erythroderma is a rare and usually severe autosomal recessive inflammatory ichthyosis [2].

It is usually apparent at birth and the newborn most often presents with a transparent covering that desquamates over 10 to 14 days [1]. After shedding of the membrane, the skin of infants with NBIE remains red, usually with a fine, white generalised scale. On the lower legs the scales are larger and darker [1]. The mucous membranes are often spared while palms and soles involvement is variable.

In rare instances the autosomal recessive ichthyosis is associated with storage of neutral lipid in multiple tissues, the Chanarin-Dorfman syndrome [3].

Turner's syndrome is a condition caused by numeric and structural abnormalities of the X chromosome and affects ~1 per 2500 females at birth [4]. The major features are short stature and primary ovarian failure. Short stature and monosomy for a maternal X chromosome have been implicated in impaired functionality in adult life [5]. In genetics, a mosaic or mosaicism denotes the presence of two or more populations of cells with different genotypes in one individual who has developed from a single fertilized egg [6]. In individuals with mosaic Turner's syndrome, cells with X monosomy (45,X0) may occur along with cells that are normal (46,XX), cells that have partial monosomies, or cells that have a Y chromosome (46,XY) [7]. In a person with mosaic Turner syndrome, some cells have the normal number of 46 chromosomes, but other cells are missing one X or there are structural defects in the second X. Many different karyotypes fall under the category of "mosaic". Some of the most common mosaic karyotypes are 45,X0/46,XX and 45,X0/46,XY [8].

Here we report a young girl with an unusual association of non-bullous ichthyosiform erythroderma with a mosaic form of Turner's syndrome.

### Case Report

A 31 year old unmarried lady presented to the outpatient department with complaints of generalized dryness and scaling of her

skin right from her infancy. The scaling increased gradually in a few weeks from birth. There was no history of any blister formation at any time, at birth or later. A detailed history did not reveal presence of any plastic-like membrane at birth. As a child, the patient had generalized redness and scaling of the skin of her body with some improvement after puberty. The skin of palms and soles was hyperkeratotic and there was no mucosal involvement. The patient was born to non-consanguineous parents. There was no history of any similar complaints in the family. Further history revealed that she had irregular menstrual cycles. She had attained menarche at 12 years age, but from the onset her menstrual cycles were irregular, occurring every two-three months. The flow was less and persisted for only two or three days. For this, she had been taking treatment previously off and on. However, she was amenorrhoeic since the past 3 months.

Examination revealed small, dry scales distributed over the extensors of the upper extremities, the face, scalp and the trunk. The scales were larger and hyperpigmented over the lower extremities. The palms and soles also showed scaling and hyperkeratosis. The nails were brittle and had longitudinal ridging.

A chance examination showed that the patient had a short stature and minimal webbing of the neck. Further evaluation showed she had widely spaced nipples (Tanner IV) with absent pubic and axillary hairs (Tanner I). She also had flexion deformity of the first interphalangeal joint of the little finger bilaterally (Figures 1a-1d).

With these findings a provisional diagnosis of non-bullous ichthyosiform erythroderma with Turner's syndrome was made. The patient was taken up for further evaluation. A complete blood

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count, renal and liver function tests revealed no abnormality. An echocardiography showed normal findings. An ultrasonography of the lower abdomen showed hypoplastic uterus with non visible left ovary. This was further followed up with a diagnostic laparoscopy which showed a hypoplastic uterus with normal ovaries. For confirmation a karyotyping was done which showed a mosaic form of Turner syndrome with 24% 45X0 and 76% 46XX (Figure 2). A skin biopsy from the lesion showed hyperkeratosis, mild parakeratosis and acanthosis, a normal granular layer and an irregular psoriasiform hyperplasia of the epidermis. The histopathology features were suggestive of lamellar ichthyosis (Figure 3). But since the histopathological features of lamellar ichthyosis and nonbullous ichthyosiform erythroderma are not very accurately distinguishable and clinically the features were in favour, hence a diagnosis of non-bullous ichthyosiform erythroderma with mosaic form of Turner's was made. The patient was started on

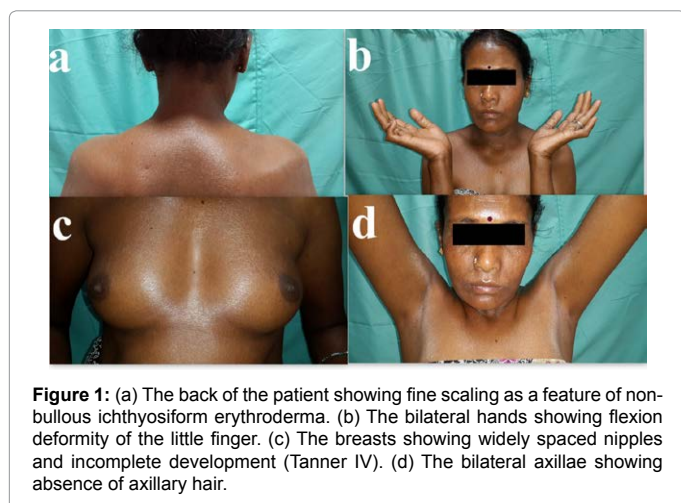
low dose isotretinoin therapy along with topical moisturizers for her ichthyosis and is on regular follow-up.

## Discussion

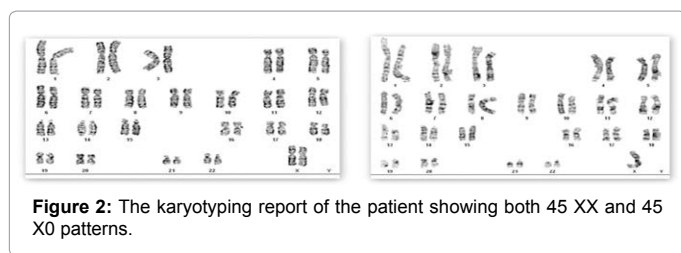
Non-bullous ichthyosiform erythroderma is usually rare and is considered a less severe variant of lamellar ichthyosis [3]. There is an expanding list of syndromes that combine with ichthyosis. Here we report an association of a mosaic Turners Syndrome (45X0/46XX) and congenital non-bullous ichthyosiform erythroderma is reported here. The karyotype of the 31 year old female revealed 24% 45X0 and 76% 46XX cell line. No such association has been reported in literature so far. A few other numerical abnormalities reported in literature are, a 16-year-old girl with x-linked ichthyosis and Turner's syndrome, a 24-year-old man with Down syndrome and prominent ichthyosiform skin and a Klinefelter Syndrome with ichthyosis vulgaris and pulmonary stenosis [9-11]. The case is reported for its rarity and for being the first of its kind to be reported in literature.

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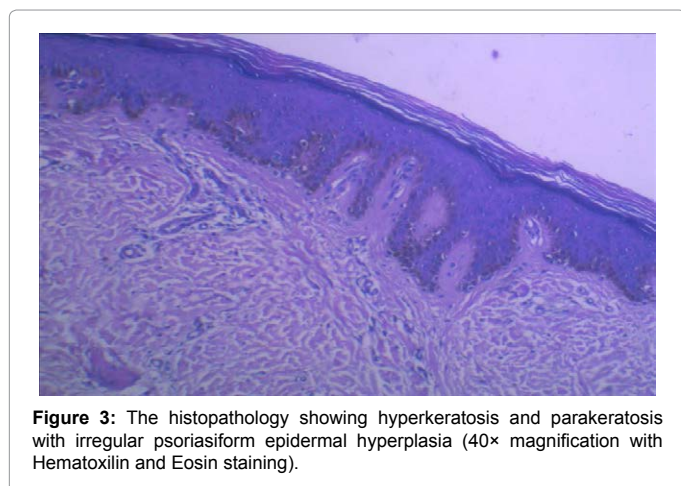
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**Figure 1:** (a) The back of the patient showing fine scaling as a feature of non-bullous ichthyosiform erythroderma. (b) The bilateral hands showing flexion deformity of the little finger. (c) The breasts showing widely spaced nipples and incomplete development (Tanner IV). (d) The bilateral axillae showing absence of axillary hair.



**Figure 2:** The karyotyping report of the patient showing both 45 XX and 45 X0 patterns.



**Figure 3:** The histopathology showing hyperkeratosis and parakeratosis with irregular psoriasiform epidermal hyperplasia (40x magnification with Hematoxylin and Eosin staining).