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Original Research Paper

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ICHTHYOSIS PREMATURITY SYNDROME: A RARE CASE REPORT

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ABSTRACT Ichthyosis prematurity syndrome (IPS) is a rare autosomal recessive disorder of cornification characterized by the clinical triad of premature birth, thick caseous desquamating epidermis, and neonatal asphyxia. It is caused by mutations in the SLC27A4 gene or FATP4 gene encoding fatty acid transport protein 4 (FATP4) leading to defective lipid homeostasis, affecting keratinocyte differentiation and skin barrier formation. The IPS babies are most commonly born premature and at gestational weeks 30 to 34. Immediately after birth asphyxia is typically noted probably due to aspiration of amniotic debris. At birth, the skin is covered by a thick, caseous, desquamating epidermis. Later in the course of the disease, the child's health improves rapidly, changing to follicular hyperkeratosis, dryness of the skin, and fine white scaling at the scalp that persists into adulthood. Overall, IPS is recognized rarely, and only a few cases have been reported. Diagnosis is essential in order to reassure parents about the benign course of the disease after complications in the perinatal period and to offer genetic counselling.

KEYWORDS : ICHTHYOSIS, CORNIFICATION, SYNDROME, PREMATURITY

INTRODUCTION

Ichthyosis prematurity syndrome (IPS) is a rare autosomal recessive disorder of cornification characterized by the clinical triad of premature birth, thick caseous desquamating epidermis, and neonatal asphyxia.[1] It is caused by mutations in the SLC27A4 gene or FATP4 gene encoding fatty acid transport protein 4 (FATP4) leading to defective lipid homeostasis, affecting keratinocyte differentiation and skin barrier formation. The estimated prevalence of IPS is 1 in 200,000 births worldwide and 1 in 50,000 in Sweden and Norway.[2] In spite of its well-described clinical features, IPS is recognized rarely, and only a few cases have been reported worldwide.[3]

CASE REPORT

A preterm female baby born at a gestational age of 34 weeks with a birth weight of 2300g. Immediately after birth she developed respiratory distress and was intubated for a short time and admitted in a neonatal care department for 7 days. Cutaneous examination of the baby revealed multiple hyperkeratotic yellowish plaques with scaling covering the scalp, face, all 4 limbs, abdomen, and back giving an impression of excessive vernix caseosa (Fig. 1). Based on clinical presentation and physical examination, the baby was clinically diagnosed as ichthyosis prematurity syndrome. The baby was treated with emollient. On subsequent follow-up after 3 months, near complete disappearance of thick hyperkeratotic plaques was noted over the extremities, abdomen and back with persistent minimal plaque and mild scaling over the forehead and scalp (Fig. 2). The parents were reassured about the condition and asked to continue the emollient and to follow up regularly.

DISCUSSION

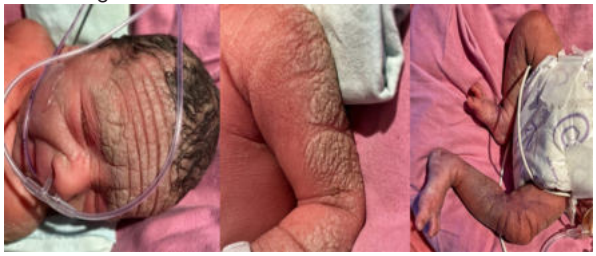
IPS is a congenital condition belonging to a heterogeneous group of autosomal recessive congenital ichthyoses.[1] It has been shown that the FATP4 gene has an essential role in the development and maintenance of the epidermal barrier. This gene is found to be widely expressed in various human tissues however it appears to be more prominent in the epidermis. It has been postulated that for the formation of the intercellular lipid matrix of the stratum corneum, the transport functions and regulation of FATP4-related lipids place a critical role. In humans, the cause of hyperkeratosis and an alteration of the skin barrier has been found to be due to the lack of formation of the lipid matrix.[2-4] In 2004, genome-wide linkage analysis was conducted by a Scandinavian group in 16 families with IPS from Norway and Sweden. The IPS locus is located on chromosome 9q34.[5]

In IPS, the pregnancy is complicated by polyhydramnios and the shedding of large amounts of epidermal cells leads to opaque amniotic fluid (starry-sky appearance on ultrasound).[6] The IPS babies are most commonly born premature and at gestational weeks 30 to 34. Immediately after birth asphyxia is typically noted probably due to aspiration of amniotic debris. At birth, the skin is covered by a thick, caseous, desquamating epidermis. Later in the course of the disease, the child's health improves rapidly, changing to follicular hyperkeratosis, dryness of the skin, and fine white scaling at the scalp that persists into adulthood.[7]

Frequent hyper eosinophilia is noted in IPS at birth. Biopsy shows non-specific signs of hyperkeratosis, acanthosis, and inflammation of the dermis. Curved trilamellar membranes in the stratum corneum and increased lipid vacuoles are seen in electron microscopy.[8]

Atopic manifestations including atopic dermatitis, asthma, and allergic rhinitis are seen in a majority of the children with IPS and high titres of IgE are noted rarely.[9]

Overall, IPS is recognized rarely, and only a few cases have been reported. Diagnosis is essential in order to reassure parents about the benign course of the disease after complications in the perinatal period and to offer genetic counseling.



(a) (b) (c)

Figure 1 At birth: (a) Thick vernix caseosa-like scale on the face and scalp (b) Cobblestone ichthyosis over the upper limb (c) Yellowish hyperkeratotic plaques with scaling over lower limbs.

(a) (b) (c)

Figure 2 At 3 months: (a) Minimal plaque with mild scaling on the forehead and scalp (b) Near complete disappearance of the lesions over upper limb and (c) Lower limbs.

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