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Ichthyosis Prematurity Syndrome: A Rare But Easily Recognizable Ichthyosis

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Abbreviations:IPS:

Ichthyosis prematurity syndrome; SLC27A4: solute carrier family 27 member 4; FATP4: fatty acid transport protein 4; NICU: Neonatal Intensive Care Unit

Keywords:

Ichthyosis prematurity syndrome; SLC27A4, verruciform hyperkeratotic; plaques; cobblestone; premature birth; neonatal asphyxia

1. Introduction

Ichthyosis prematurity syndrome [IPS, OMIM608649] is a rare autosomal recessive disorder characterized by the clinical triad of premature birth, ichthyosis, and neonatal asphyxia [1]. It is caused by a mutation in SLC27A4 (solute carrier family 27 member 4, which encodes fatty acid transport protein 4 [FATP4]) [2].

Here, we describe an IPS patient with homozygous pathogenic mutation at the initiation codon site (c.1>G, p. Met1Val) in SLC27A4 gene to raise awareness of this rare syndrome despite its distinctive features as we believe it is still under diagnosed.

2. Case Report

A 33-week gestation baby boy, born for a healthy consanguineous parent by caesarian section due to premature rupture of membrane and chorioamnionitis. At birth, he weighed 2.7 kg (10th centile), 47.5cm in length (10th centile) and his head circumference was 34 cm (10th centile). His Apgar scores were 7 and 8 at 1st and 5th minute and he was admitted to Neonatal Intensive Care Unit (NICU) due to respiratory distress and was intubated on continuous positive pressure mechanical ventilation. At 32 weeks of gestation, antenatal course of dexamethasone was given to promote lung maturity.

On examination, the baby had no dysmorphic features. His skin examination revealed generalized thick verruciform hyperkeratotic plaques with cobblestone appearance covering all of his body including the scalp with focal areas of hair loss (Figure 1). Mucous membranes and nails were normal. Ophthalmology examination was normal. His blood eosinophilia at birth was 1700 cell/mm3, peaked at day 7 reaching 2300 cell/mm^3 and normalized at the age of 1 month. The clinical diagnosis of IPS was rendered and later proven by whole exome sequencing revealing that he was carrying a homozygous pathogenic mutation (c.1A>G, p. Met1Val) in the SLC27A4 gene with parents heterozygous for the same mutation.

The proband was managed by incubator with 80% humidity, in-

travenous hydrocortisone at a dose of 1mg/kg/dose 6 hours for 7 days. Skin was managed conservatively by applying frequent, generous amount of petroleum jelly. Over the next 4 weeks, ichthyosis resolved gradually and completely (Figure 2). The patient developed recurrent pneumonia with recurrent admission to the hospital and was treated with intravenous antibiotics.

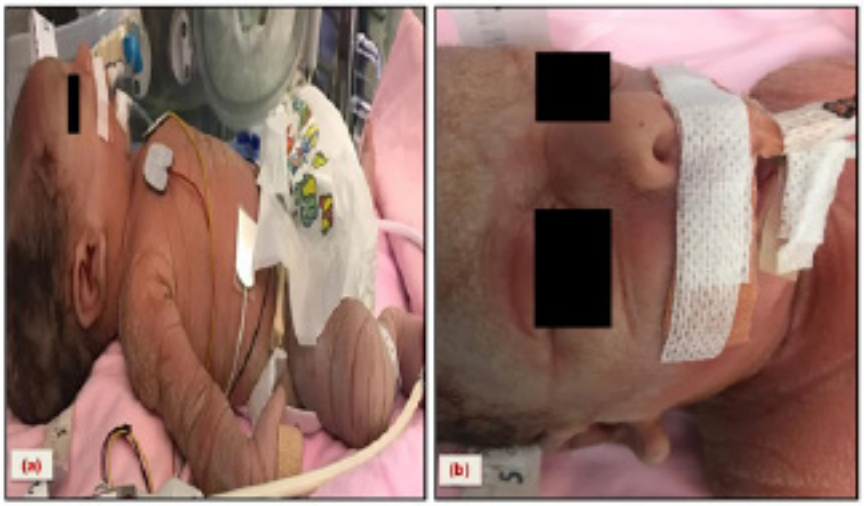

Figure 1: Photograph of the patient at birth.
A) Multiple thick verruciform hyperkeratotic plaques with cobblestone appearance covering all of his body including the scalp.
B) Cobblestone plaques covering the face, more prominent over the forehead.

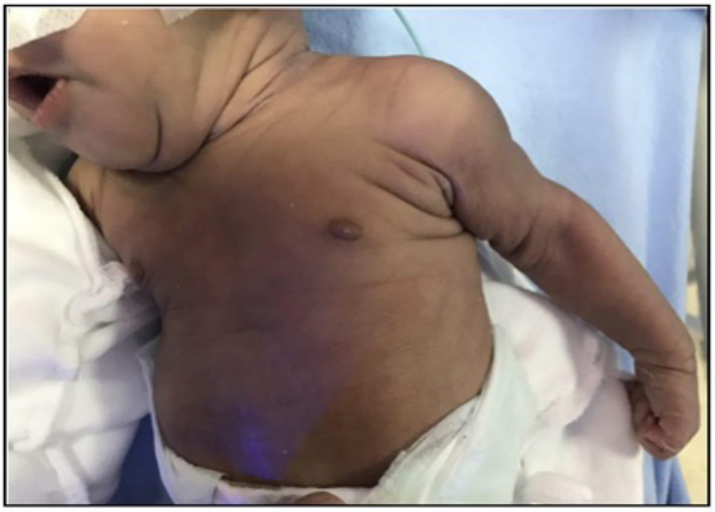

Figure 2: Photograph of the patient at the age of 1 month showing resolution of ichthyosis.

3. Discussion

Ichthyosis prematurity syndrome is a rare autosomal recessive disorder, first recognized in 1993 [3]. Leaute-Labreze et. al, suggested the term "Self-Healing Congenital Verruciform Hyperkeratosis" [4], because skin findings improve spontaneously unlike other forms of congenital ichthyosis, although some patients may persist with skin xerosis or atopy [5]. Neonatal asphyxia is thought to be due to aspiration of skin debris that shed into amniotic fluid1. Antenatal ultrasound may show separation of chorionic and amniotic membranes, polyhydramnios with starry sky appearance [4,5]. Histopathology of skin is pathognomonic with acanthosis, hyperkeratosis and characteristic aggregates of curved lamellar structures in the perinuclear cytosol of stratum corneum and stratum granulosum [6]. Perivascular inflammation with eosinophilia was seen in some cases [6].

On electron microscopy, some authors described it as worm-like structures in corneocytes4. Transient blood eosinophilia like in our patient have been noticed occasionally [1].

Respiratory complications are the leading cause of death, due to inhalation of debris [1]. Considering that lung pathology might be

due to aspiring skin debris, we believe that systemic steroids might help mitigating the respiratory complications [7].

Ichthyosis prematurity syndrome was previously reported due to mutation within SLC27A4 that encoding FATP4[2]. The described c.1A>G, p.Met1Val mutation in our patient was previously reported resulting in loss of initiation codon leading to pathological effect on FATP4 protein [8]. The FATP family are transmembrane proteins that transport exogenous fatty acids into cells and activate them. They also function as acyl-CoA synthetases with specificity for very long-chain fatty acids (VLCFA), reducing VLCFA-CoA synthetase activity and incorporation of VLCFA into neutral and polar lipids [2].

Animal studies suggest that multiple FATP specifically FATP4 are important for skin barrier function, particularly during embryonic and neonatal period, but are not vital postnatally as other FTAP may compensate [9]. This may explain spontaneous skin improvement soon after birth [2].

In conclusion, we are presenting and emphasizing the pathognomonic and special cobblestone appearance of ichthyosis in IPS. We also think that systemic steroids may improve the prognosis.

4. Acknowledgement

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