

A Rare Case of Nonbullous Congenital Ichthyosiform Erythroderma in a Preterm Neonate - A Case Report

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Abstract: *Nonbullous congenital ichthyosiform erythroderma (NBCIE) is a rare non blistering disorder and one form of severe congenital ichthyosis. The incidence is 1 in 300,000 individuals of all races. An affected newborn is usually born as a collodion baby. It presents with fine grey white scales after shedding of the collodion membrane. Here we report one such rare case of NBCIE, diagnosed in a preterm neonate at 3 weeks of age with consistent clinical and skin biopsy findings.*

Keywords: Nonbullous congenital Ichthyosiform erythroderma, Collodion baby

1. Introduction

Nonbullous congenital ichthyosiform erythroderma (NBCIE) is an extremely rare skin disorder characterised by abnormal scaling of skin with generalised erythroderma. The pattern of inheritance is autosomal recessive. The disorder affects 1 in 300,000 individuals of all races (1). There is an inherited abnormality that affects normal skin shedding. The genes involved in mutation are TGM1, ALOX12B, ALOXE3 and ichthyin. It is not contagious and not associated with any internal abnormalities.

2. Case Report

A male neonate weighing 1.8kg was born at 36 weeks of gestation by caesarean section at Chengalpattu Medical College hospital, indication being fetal distress with severe oligohydramnios. The mother was a known case of pregnancy induced hypertension on antihypertensive medication. Baby sustained perinatal asphyxia at birth, was apneic with HR < 100. The neonate was resuscitated and required endotracheal intubation and thereafter admitted in neonatal intensive care unit. On physical examination there was no facial dysmorphism and no external congenital malformations. Baby's skin was found to have a rough texture. Dermatologist opinion was obtained, was diagnosed as a form of ichthyosis, advised external application of liquid paraffin and a review after 2 weeks. There was no family history of hereditary skin diseases. Baby was managed with mechanical ventilation for 2 days and oxygen support gradually tapered over a period of 7 days. There were no seizures during hospital admission. The lab investigations were within normal limits, vitals stabilised, general condition improved, discharged and advised follow up.

Baby was again readmitted at day 24 of life, for pneumonia associated respiratory failure. The initial stabilisation was done and this time, examination of the skin of the neonate revealed generalised erythema, with a rough texture. There was peeling of skin with white scales all over the body. The

neonate had narrow palpebral fissure and absence of eyebrows. There was no ectropion or eclabium. There were no contractures and no restriction of joint mobility. The external genitalia was normal. The mucous membranes were spared. (fig 1,2,3) Basic investigations were under normal range. Septic screening was negative. Thyroid profile was normal. Skin biopsy showed stratified squamous epithelium with vacuolation of upper and midspinous layer; hyperkeratosis with large keratohyaline granules in the vacuolated granulosa cell layer. The neonate was diagnosed as a case of nonbullous congenital ichthyosiform erythroderma. Hydration was maintained and external application of emollients were used. Despite intensive care, baby succumbed to cardiorespiratory failure, due to underlying medical illness.

3. Discussion

Nonbullous congenital ichthyosiform erythroderma is a rare inherited skin disorder. The affected newborn is initially covered with a parchment like membrane-collodion (2). After shedding of this membrane, generalised erythroderma and fine white scales become evident. Other features include alopecia, skin fissures, nail dystrophy but the mucosal surfaces are spared (3). Palmoplantar hyperkeratosis is seen in 70% of cases. Involvement of the face is characterised by flattening of ears, nose, ectropion, etc. Hypohidrosis and heat intolerance, occur due to obstruction of sweat ducts and pores (4).

Histopathological examination reveals compact hyperkeratosis and increase in thickness of stratum corneum. There are variable mild parakeratosis, acanthosis, hypergranulosis and accentuation of rete ridges. The close differential diagnosis is lamellar ichthyosis where there are large dark scales compared to the fine white scales of NBCIE (5).

This disorder is also associated with certain syndromes like Sjogren-Larsson syndrome, Dorfman - Chanarin syndrome, Neu-Laxova syndrome etc. Our case did not have any

syndromic features and there was no collodion membrane at birth. The neonate had a rough texture of skin at birth with the erythema and scales being evident after 3 weeks of life. The typical clinical features and suggestive skin biopsy findings confirmed the diagnosis of Nonbullous congenital ichthyosiform erythroderma.

There is no specific marker for prenatal diagnosis. Genetic counselling is of prime importance. Treatment is only symptomatic aiming at hydration, lubrication and keratolysis. Emollients remain the mainstay of treatment. Prevention of sunstroke and sunburn is important.

4. Conclusion

Nonbullous congenital ichthyosiform erythroderma is a rare condition that presents in the neonatal period. In our case the diagnosis was evident at 3 weeks of age after shedding of the initial parchment membrane. Although fetal skin biopsy with amniocyte pelleting at 17-22 weeks, looking for premature or abnormal keratinisation is possible, the results are unsatisfactory due to phenotypic heterogeneity and the need for multiple biopsies. This case is reported for its rarity and for the classical clinical and histopathological diagnosis of Nonbullous congenital ichthyosiform erythroderma.

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Figure 1&2: Rough dry skin with scales



Figure 3: loss of eyebrows