



## Case Report

### Congenital Ichthyosis – A Case Report

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#### ABSTRACT

Disorders of cornification (ichthyoses) are a primary group of inherited conditions characterized clinically by scaling and histopathologically by hyperkeratosis. Collodion and harlequin babies are types of these disorders. These disorders may cause considerable disfigurement and psychological stress to caretakers. Early diagnosis is helpful to predict prognosis and to provide supportive management for the patients and families. According to Various case reports the mortality of collodion babies varies from 9 to 45% with the highest mortality rates seen in harlequin ichthyosis. This is case report of a collodion baby (congenital ichthyosis) admitted in NICU of Grant Medical college and JJ hospital. The baby was born at a peripheral hospital and was referred to JJ hospital for NICU care. He was managed according to standard protocol. Strict asepsis and thermoregulation was maintained. Oxygen supplementation, IV fluids and IV antibiotics were given. Local application of emollient and eye care was provided. The baby got stabilized and was started initially on orogastric tube feeding and later was shifted to breastfeeding and was subsequently discharged after establishment of breastfeeding.

**KEYWORDS:** Ichthyosis, Collodion baby.

#### INTRODUCTION

Collodion baby is a rare genodermatosis of unspecified inheritance, affecting both sexes equally. Less than 300 cases of collodion babies have been reported in the literature since 1892 when the term was first used by hallopeau and watelet[1]. The term collodion baby refers to the babies who are covered at birth by a thick, taut membrane resembling oiled parchment or collodion, which is subsequently shed [2]. Its peculiar phenotype is also sometimes called 'Dipped in the wax' or 'sausage skin' appearance [3].

The most important clinical data concerning collodion baby is the presence of disseminated ichthyosiform genodermatosis characterized by dry skin, scaling, generalized erythroderma and hyperkeratosis, reminiscent of fish scales. This type of dermatosis is also known by the generic name of ichthyosis. Collodion Baby' is not a disease entity but the common representation of several well-defined disorders which can be distinguished by reference to family history, and by observing the natural history of the condition [4]. Neonatal complications can occur in 45% of all collodion babies, leading to a mortality rate of ~11% in the first few weeks of life [5]. The mortality and morbidity is far more in harlequin ichthyosis which is the most severe form of collodion baby (harlequin ichthyosis) [6].

The babies with harlequin ichthyosis usually succumb to sepsis and fluid loss in neonatal period but rarely the child may survive up to several months or even years [7]. Harlequin babies are at high risk for hypo/hyperthermia, dehydration, respiratory distress, hypoventilation, malnutrition, hypernatremia, seizure, and skin infection [8].

#### CASE REPORT

A one-day-old, preterm, female baby, born to non-consanguineous parents, weighing 1.9 kg was referred from a private practitioner for abnormal appearance of skin and also for NICU care. Baby was delivered by Normal Vaginal Delivery at a peripheral hospital. Baby cried immediately after birth but developed respiratory distress in the form of tachypnea and intercostal and subcostal retractions. On admission the baby was having respiratory distress in the form of grunting and subcostal and intercostal retractions. Also there was presence of thick skin with fissuring at multiple areas of skin, general hyperkeratinization, small and flattened nares, open mouth, flat fontanel, ectropion, immature eyes and auricles, eclabium and generalized flexed attitude of limbs (Figure 1).

**Figure1: Ichthyosis (collodion Baby)-Hyperkeratinization, open mouth, ecteropion and flexed posture is visible**



**Figure 2 : Same Baby with marked improvement at the time of discharge.**



Baby was admitted in NICU. A chest Xray was done which was normal. She was started on iv fluids and iv antibiotics. Oxygen supplementation by hood was given. Frequent application of emollients was also started. Lubricant eye drops were given to prevent drying and subsequent corneal involvement. Strict thermoregulation and asepsis was maintained. Blood gas analysis was within normal limits. Initial septic screen was normal. Other biochemical markers like serum calcium, liver and kidney function tests and electrolytes were all within normal limits. An

ophthalmologic consultation was done for ecteropion. The respiratory distress settled down on Day 2 of life. The membrane started peeling off from day 3, peeled skin left erythematous surface underneath. On day 5 of admission baby was haemodynamically stable and IG feeds with expressed breast milk was started (Fig-2)The baby tolerated feeds well which were gradually increased to full feeds. Later baby was shifted to direct breast feeding and was discharged on day 12 of life.

## DISCUSSION

Disorders of cornification (ichthyoses) are a primary group of inherited conditions characterized clinically by pattern of scaling and histopathologically by hyperkeratosis. These consist of spectrum of conditions with different phenotypes and severity. Majority of collodion babies develop autosomal recessive ichthyosis either lamellar or congenital ichthyosiform erythroderma [9]. They Within some days after birth baby reveals extensive scaling of the skin. As the age progresses the scaling will be concentrated around the joints in areas such as the groin, axillae, and the inside of elbow and neck. The skin layer peels off itself gradually. The babies suffering from these disorders are prone for developing hypo/hyperthermia, sepsis and dehydration. The practical consequences for paediatric management are manifold. A collodion baby is the equivalent of a highly premature baby in terms of epidermal barrier, with a high transepidermal water loss and a major risk of dehydration and hypothermia [10].

This transepidermal water loss can be 6 to 7 times higher than normal skin [11]. The management is mainly supportive. Collodion babies needs to be nursed in a humidified incubator after borth to prevent dehydration and hypothermia. Also strict asepsis is a must. Liberal use of emollient is necessary. Harlequin ichthyosis is severe form of this spectrum of disorders. Inheritance is autosomal recessive. Common morphologic abnormalities include hyperkeratosis, accumulation of lipid droplets within corneocytes and absence of normal lamellar granules. One type has an altered catalytic subunit of 2A protein phosphorylase which is encoded on chromosome 11 [9].

Harlequin babies may have respiratory failure as a result of restricted chest expansion and skeletal deformities. Feeding problems may result in low blood sugar, dehydration, and kidney failure. In addition, temperature instability and infection would be common. The mortality of harlequin babies is high and most of the victims die within a few weeks of birth because of secondary complications such as infection and dehydration [12].

## CONCLUSION

Collodion or harlequin babies belong to same group of disorders namely disorders of keratinization (ichthyosis). The peculiar features make the diagnosis obvious. The management is essentially supportive. Strict asepsis, thermoregulation, use of emollients for skin and lubricant drops for eyes may prevent further complications. Extra fluid loss needs to be replaced. After stabilization hearing may be tested to rule out KID (keratitis, ichthyosis and deafness) syndrome.

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