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COLLODION BABY SYNDROME: A CASE REPORT FROM PIMS HOSPITAL, ISLAMABAD

Um ul Baneen Zehra[1], Fateema Tanveer[2], Khadeejah Sajwani[3], Mishkat Aslam[1], Mehar Zahra[1], Hamyel Tahir[1], Maham Nadeem[1], Sadia Rekhum[4], Tayyaba Maheen[2], Mohammad Shah Hussain[1], Muhammad bin Aslam[5]

[1]Combined Military Hospital Lahore Medical College and Institute of Dentistry
[2]National University of Sciences and Technology (NUST), Institute of Health Sciences
[3]Manchester University National Health Service (NHS) Foundation Trust
[4]Rahbar Medical & Dental College, Lahore
[5]Continental Medical College, Lahore

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Corresponding Author:
Um ul Baneen Zehra, Combined Military Hospital Lahore Medical College and Institute of Dentistry

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ABSTRACT

Collodion Baby Syndrome (CBS) represents a distinctive and uncommon congenital disorder characterized by a striking clinical presentation of collodion membrane upon birth. Collodion baby, also known as 'collodion fetus,' is frequently observed in various forms of congenital ichthyoses, many of which follow an autosomal recessive pattern of inheritance. These conditions include lamellar ichthyosis (LI), congenital ichthyosiform erythroderma (CIE), and self-healing collodion baby.[1] This case report describes the intricate dermatological challenges encountered in the management of a neonate diagnosed with CBS, emphasising the nuanced complexities that clinicians must confront in providing optimal care. A premature female infant was born at 35 weeks via emergency cesarean section who presented with a parchment-like membrane covering her entire body, which is a typical hallmark of CBS. She was taken to the Neonatal Intensive Care Unit where a multidisciplinary management approach encompassed meticulous skin care and assessment of transcutaneous water loss to optimize skin hydration. Vigilant monitoring of electrolyte imbalances as well as the potential complications of infection and fluid imbalance were essential components of the comprehensive care plan. Despite all efforts, the baby died on the fourth day of life due to a severely compromised epidermal barrier and its complications. In cases such as these, prompt identification and treatment is vital.

Introduction

The term collodion baby (CB) describes a newborn whose entire body is enveloped in a thin, flexible, parchment-like membrane. This condition commonly presents with ectropion, eclabium, hypotrichosis, underdeveloped nasal and ear cartilage, and pseudocontractures. Genetic analysis revealed that the child carried a homozygous mutation in the *ALOX12B* gene, resulting in the loss of a glutamine amino acid at position 136 of exon 3, while the parents were healthy heterozygous carriers of this mutation.[3] Collodion baby represents an exceptionally rare dermatological emergency, with an estimated incidence ranging from 1 in 50,000 to 1 in 100,000 births.[7]

Collodion babies are likely underreported, especially when cases present mildly. Our encounter with such a case emphasized the significance of diligent skin care, cautious use of products, and strict adherence to cleanliness when tending to an infant with this syndrome. Given the rarity and diverse manifestations of the disease, individual clinicians may lack extensive experience, highlighting the need for a standardized protocol to address the evolving nature of this condition.

Case

A female neonate, weighing 2.1 kg was delivered via emergency LSCS at 35 weeks of gestation on 12/03/23 at 12:45 am due to preterm labor and a non-reactive CTG. On examination, her length and head circumference were 45 cm and 26 cm, respectively. She was vitally stable with a respiratory rate of 38 breaths per minute, a heart rate of 136 beats per minute, and had an immediate cry after birth. Apgar's score was 8/9 at 1 and 10 minutes. A thorough examination revealed that she had very hard skin which consisted of stiff, flat membranes with fissures covering the whole body along with generalized edema. Bilateral distortion was observed in the pinna along with parchment-like skin over the ears. She had bilateral ectropion with no visible pupils and sclera (Figures 1 and 2). A systemic examination revealed no abnormalities and no other obvious anomalies were identified.

Family and antenatal history was obtained from the mother, according to which the mother was 21 years old, with an O-positive blood group. She had no history of pregnancy-induced hypertension or gestational diabetes mellitus. The parents were non-consanguineous. A pedigree chart was also made, and there was no positive family history for any dermatological or genetic diseases. There was no history of any complications during the pregnancy, and the anomaly scan did not reveal any anatomical abnormalities. The baby was diagnosed as a case of collodion baby syndrome.

The neonate was immediately shifted to the Nursery and managed in an incubator. She was closely monitored and fed every 2 hours via an NG tube, and bowel movements were also normal. She was given oral antibiotics with nasogastric feed, Polyfax ointment (topical emollient) for skin, Tear Plus eye drops (artificial tears) for lubrication, and was covered with paraffin gauzes, which led to subsequent improvement in skin condition. Despite all possible treatment, on 16/03/23, her fourth day of life, she suddenly collapsed. Her skin was pale, cold, and clammy, with no heartbeat. Parents were informed, and death was declared.

We obtained written consent from the mother to capture photographs and to present this case, as well as to utilize the images for academic purposes.

Fig 1:At the time of birth

Fig 2: Few hours later

DISCUSSION

Collodion baby syndrome is a rare genetic disorder characterized by the presence of a tight, shiny, translucent membrane covering the newborn's body at birth. This condition poses significant challenges for affected infants and their families due to associated complications and the potential for long-term sequelae.[1]

One of the primary challenges in managing Collodion baby syndrome is the potential for skin barrier dysfunction, which predisposes affected infants to infections, fluid loss, and temperature dysregulation. The collodion membrane, although protective initially, can become a hindrance once it starts to crack and peel, leaving the underlying skin vulnerable. Therefore, meticulous skin care and hydration are essential components of management to prevent complications and promote skin integrity.[2]

Furthermore, Collodion baby syndrome often serves as a clinical marker for underlying genetic mutations affecting skin development and function. Mutations in genes such as *transglutaminase 1* (TGM1) have been implicated in the pathogenesis of the condition, disrupting the normal process of epidermal differentiation and cornification. Understanding the genetic basis of Collodion baby syndrome not only aids in diagnosis but also has implications for genetic counseling and family planning, as affected individuals may have an increased risk of passing on the condition to their offspring.[3]

It is an uncommon condition characterized by significant morbidity and mortality during the neonatal phase. These infants are often premature and have a parchment-like membrane that sheds over time, revealing an underlying skin disorder. Approximately one-tenth of these cases may see a transition to normal skin. This condition, known as 'self-healing' collodion baby, represents one end of the spectrum, while the severe Harlequin fetus represents the other end.[6] The latter often leads to death within days or weeks due to respiratory distress, feeding difficulties, and severe skin infections. Survivors may experience severe ichthyosis and neurological issues. The barrier function of the skin is compromised once the collodion layer dries, making the baby vulnerable to infections, fluid loss, and temperature instability. Movement restrictions from the thick membrane can hinder breastfeeding and cause breathing problems. The primary pathology is disordered cornification, leading to collodion membrane formation, with management typically requiring dermatological and pediatric expertise. Treatment involves maintaining a humidified, neutral-temperature environment, providing supportive care such as IV fluids and tube feeding, and avoiding debridement of the collodion membrane. Emollients may increase infection risk in some cases.[4,6]

Management of collodion baby syndrome involves a multidisciplinary approach, including a dermatologist and pediatric team. The infant is initially kept in a humidified, neutral-temperature environment like an incubator. Supportive treatments such as intravenous fluids and tube feeding may be necessary to maintain hydration and nutrition. The skin should be kept soft, and scaling should be minimized, though the collodion membrane must not be debrided. Emollients like petroleum jelly are recommended to avoid infection risks, though some studies caution against other

emollients due to potential complications, such as increased infection risk and intoxication from topical products. Acitretin (1 mg/kg/day) has been shown to be effective and safe for managing congenital ichthyosis. For severe ectropion, artificial tears may be required, and mild topical steroids can help reduce secondary inflammation.[2]

In addition to the physical challenges associated with Collodion baby syndrome, affected infants and their families may also face psychosocial burdens. The visible nature of the condition can lead to stigmatization and emotional distress for both the affected child and their caregivers. Therefore, comprehensive care for Collodion baby syndrome should include psychosocial support and counseling to address the emotional needs of affected individuals and promote resilience within the family unit.[5]

Despite the significant morbidity associated with Collodion baby syndrome, there is variability in outcomes among affected individuals. While some infants may experience spontaneous resolution of the collodion membrane and go on to lead relatively normal lives, others may develop long-term complications such as severe ichthyosis and associated neurological impairments. Therefore, prognostication in Collodion baby syndrome requires careful consideration of individual clinical characteristics and genetic factors.[6]

Additionally, genetic counseling is paramount in collodion infant cases. Parents need to be educated about the hereditary nature of the condition, the mode of inheritance (usually autosomal recessive), and the potential risk of recurrence in future pregnancies. They should also be made aware of available prenatal diagnostic tests such as chorionic villus sampling or amniocentesis, which can help identify affected fetuses in subsequent pregnancies, enabling informed decision-making regarding management and future family planning.[8]

Moving forward, research efforts aimed at elucidating the underlying genetic mechanisms of Collodion baby syndrome and developing targeted therapeutic interventions are warranted. Additionally, raising awareness among healthcare providers about the clinical features and management strategies for Collodion baby syndrome is crucial to ensure timely diagnosis and optimal care for affected individuals. By addressing the multifaceted challenges associated with Collodion baby syndrome, we can improve outcomes and quality of life for affected individuals and their families.

However, it's essential to note that Collodion baby syndrome is a rare condition worldwide, including Pakistan. Information on rare diseases like this may be limited due to factors such as lack of awareness, limited research, and challenges in diagnosis and reporting.

In Pakistan, healthcare professionals, particularly dermatologists and pediatricians, would likely encounter Collodion baby syndrome cases sporadically. They may rely on international literature, case reports, and collaborative efforts with specialists in other countries for guidance on diagnosis, management, and treatment options.

CONCLUSION

Collodion Baby Syndrome is a rare congenital disorder with distinctive clinical and histological characteristics. The difficulties in detecting and treating the ailment are highlighted in this case study, along with the value of interdisciplinary treatment, comfort measures, and genetic counseling. In order to further understand the underlying genetic pathways and develop new therapy modalities for patients with Collodion Baby Syndrome, further research is crucial to elucidate the genetic mechanisms and improve therapeutic interventions.

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