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Original Research Paper

Paediatrics

COLLODION ICHTHYOSIS WITH TRISOMY – 21: A CASE REPORT

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ABSTRACT

Collodion Ichthyosis is a rare disorder presenting at birth with visible scaling/ hyperkeartosis of the skin. The severity of ichthyosis and expression of associated features may be variable. The ichthyosis is known as Mendelian Disorder of cornification (MeDOC). Inborn types have different genetic background like Autosomal Dominant (AD), Autosomal Recessive (AR) and X – linked inheritance. [1] Collodion baby is a very rare form of skin disorder with an estimated incidence of 1 in 50,000 to 100,000 birth. [2] The mortality rate is around 11% in early life due to Dehydration, Electrolyte Imbalance, Sepsis because of severe skin damage. [3, 4]

KEYWORDS : Collodion baby, Ichthyosis, Trisomy – 21, Sepsis.

CASE REPORT:

I present a collodion male baby whose karyotype study revealed 47XY + 21. 25 years old mother delivered the boy baby [fig. 1] at 36 weeks gestational age via normal vaginal delivery at a private nursing home. His birth weight was 2.4 kg and the baby cried immediately after birth. The parents are in consanguinity and they have one healthy child.

Fig. 1: Baby on day 2

The neonate was brought at my OPD for dry, thick and scalded skin with deep fissures associated with feeding difficulties. Meconium and urine was passed within 24 hrs of birth. He had wide open mouth, absent eye-brows and eye-lashes with sparse scalp hair. My clinical diagnosis was Collodion Ichthyosis. The parents were counseled regarding the outcome and the baby was referred to the tertiary care hospital for NICU management. The baby was treated on the humified incubator with intravenous fluid, antibiotic and skin-care with moisturizer, topical keratolytic agent as suggested by Dermatologist. Karyotyping was done and it was found that the child have Trisomy – 21. The mother was instructed for proper skin – care and regular follower. The baby is now 8 months old having seborrhea of scalp and erythematus dry scaly skin. [fig. 2, 3]

Fig. 2: 8 Months Old

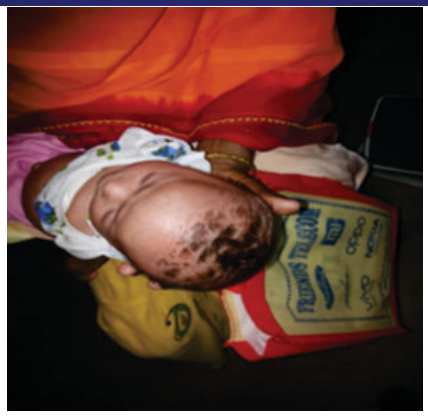

Fig. 3: 8 months old

In this study I reported collodion baby with Down Syndrome (Trisomy – 21). Skin manifestation with Trisomy – 21 are soft and velvety skin in early childhood and dry skin, secondary lichenification, ichthyosiform changes in late childhood. [5, 6] Extensive literature search I did not find Collodion Ichthyosis with Trisomy – 21.

DISCUSSION:

Hallopeau in 1884 coined the term "Collodian Baby". [7] Collodian baby is a spectrum of disorder which include Autosomal recessive congenital ichthyosis, lamellar ichthyosis, harlequin ichthyosis (most severe form) or less commonly self healing. In 20% - 25% collodian baby evolves into normal child with mild scaling in later life. [8] There is a scoring system to assess the outcome. [9] Autosomal recessive inheritance with consanguinity is seen in most of the cases of collodion baby syndrome but exact cause is unknown. [10] Harlequin ichthyosis is rare and associated with poor outcome. Trans – epidermal water loss in collodion babies are 6 – 7 times higher than normal skin's baby. [11] The baby was initially managed in a humidified incubator and skin care with emollients.

CONCLUSION:

Despite a rare disease and limited knowledge survival may be improved by providing basic care and supportive management. As a non – communicable disease psychological components like depression, anxiety and suicidal ideation involving both the patient and the parents to be managed properly.

Confilct Of Interest:

No conflict of interest.

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