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Case Report

A rare association of collodion baby with hypothyroidism from tribal area of Jharkhand: a case report

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ABSTRACT

Collodion baby is a rare congenital disorder that resembles the Harlequin foetus but is milder in severity. Although it has been reported that harlequin foetuses are associated with various abnormalities like kidney abnormalities, polydactyly, thymic atrophy, malignant keratoma, and thyroid aplasia, there are few reports of collodion babies associated with congenital abnormalities and/or disease in the literature. The case of a newborn infant admitted with skin shedding and diagnosed with collodion baby concomitant with congenital hypothyroidism is presented here. To the best of our knowledge, this is the first case report of a collodion baby with congenital hypothyroidism in the literature from the tribal region of Jharkhand.

Keywords: Ichthyosis, Collodion baby, Hypothyroidism

INTRODUCTION

Collodion baby is a Mendelian cornification (ichthyoses) disorder that is extremely rare. These are primary inherited conditions that are clinically distinguished by an abnormal scaling pattern and hyperkeratosis. It is a very rare condition that affects 1 in 50000 to 1 in 100000 live births.[1] The collodion babies are wrapped in an oiled parchment-like membrane that mimics the presence of cellophane. Flattening of the ears and nose, as well as fixation of the lips in an O shape, may be observed. Later, the membrane cracks and begins to desquamate. The collodion membrane sheds off within 2-4 weeks of birth, revealing the underlying skin disorder.

CASE REPORT

A full-term male neonate delivered by lower segment caesarean section (LSCS) to a multigravida mother. There was no history of consanguineous marriage. The newborn has a birth weight of 2.9 kg with length and head circumference being 51 cm and 34 cm respectively. At the time of birth, the vitals were normal and the APGAR score were 7 and 8 at 1, 5 min respectively as per document and had cried immediately after birth. As per the mother, the newborn body was covered with a thin cellophane like shiny membrane. There was eversion of eye lids, slight restricted movement of limbs, mild erythema over the thoraco-abdominal area. The baby was admitted for 72 hours in some local hospital without evidence of respiratory distress. Septic workup was negative and baby was discharge. Then the baby came to pediatric OPD AIIMS Deoghar at around 12 weeks of age with complain of peeling of superficial skin with all the past history revealed. The baby was diagnosed to be a case of Lamellar Ichthyosis with decreased ectropion with improved movements of limbs. The body was still covered with broken parchment like membrane with desquamation. The baby was on expressed mother milk with present satisfactory growth with weight of 6.44 kg, Head circumference of 39 cm, length 53 cm. The systemic examination appeared to be normal with no cardiac abnormality. No other associated obvious anomalies were seen. There was no history of consanguineous marriage. There was no history of such condition reported in family members except the sibling. The 1st newborn delivered to

the mother was male who died at age of 9 months, 2nd sibling was also male who died at age of 12 months. Both the babies were suffering from similar condition and later died. 3rd one is a female who survived (presently 7 years old). 4th pregnancy was aborted. 5th child is alive (at present 2 years old). 6th child is the Index case. Thyroid hormone levels were as follows: thyroid-stimulating hormone (TSH) 98.36 IU/ml, serum total triiodothyronine (T3) 101.63 ng/dl, total thyroxine (T4) 3.22 µg/dl, and thyroglobulin 348.58 ng/ml. The mother was asked to go for genetic analysis of the condition but due to poor socio-economic condition she cannot afford the investigation. Dermatological opinion was also taken. The mother was also counselled for Prenatal diagnosis in next pregnancy by fetal skin biopsy and microscopic examination of cells in amniotic fluid if possible. Topical emollients were regularly applied. Artificial tears were advised to prevent drying of eyes. The infant was closely monitored for any sepsis. Acitretin (0.5–0.75 mg/kg per day) was given. Application of non-occlusive lubricants facilitates shedding of membrane. New membrane may form at localized area after complete shedding which takes several weeks. Consent was taken from parents for case reporting and image publication.

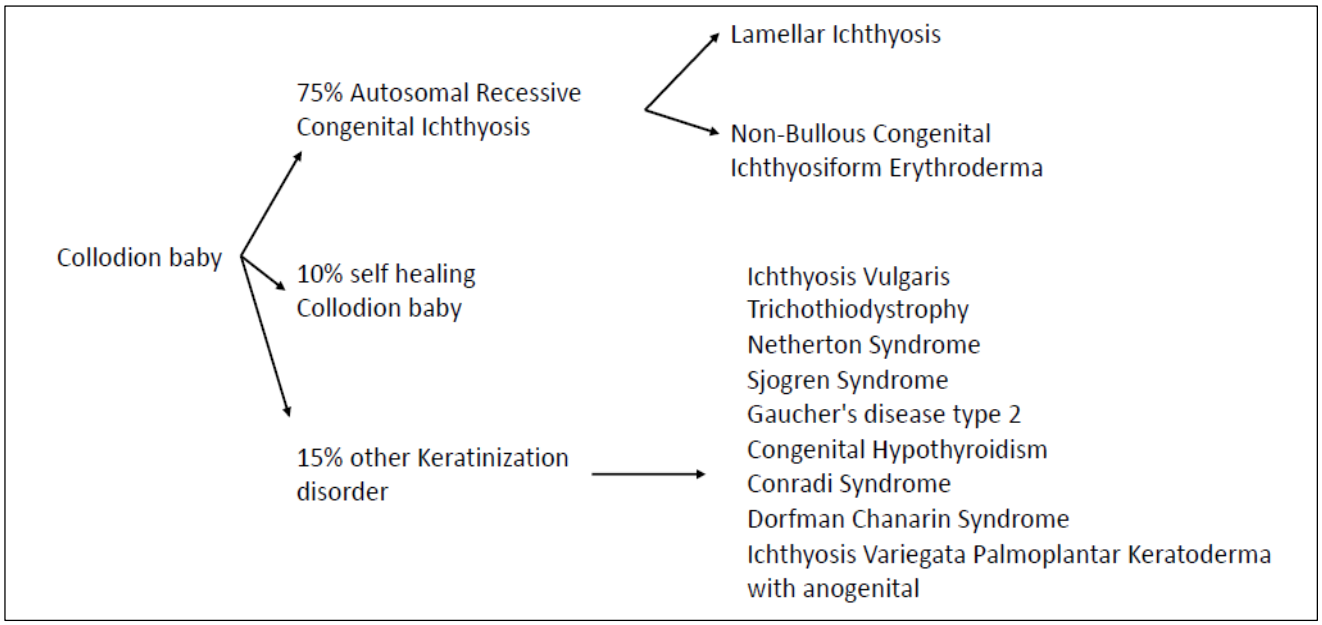

Figure 1: Outcome subtype of collodion baby.

Management and treatment

The management strategy focuses on providing protection to the skin barrier, control of infections, maintenance of fluid and electrolyte balance, and early initiation of retinoid therapy.[10] It requires the combined effort of a dermatologist, a neonatologist, and, in some cases, an ophthalmologist and an ENT specialist. The aim of treatment is to eliminate fish-like scales and reduce excessive irritation. There is also a high risk of hypernatremic dehydration in this type of patient. So it requires the patient to be placed in highly humidified incubators, and regular monitoring of the body temperature and proper nutrition are provided. An increase in the risk of infections, both cutaneous and systemic, like candida and bacterial, is possible. The first line of management is moisturising and topical keratolytic agents, as they enhance the skin barrier and facilitate desquamation. Sodium chloride, urea, vitamin E acetate, glycerol, and petroleum jelly are various agents available as moisturisers and lubricants. These infants are at increased risk of intoxication by the absorption of topical products, like salicylates or keratolytics due to impaired skin. In severe cases with marked hyperkeratosis, keratolytic agents like lactic acid, glycolic acid, salicyclic acid, N-acetyl-cystine, and glycol can be used. Ectropion is managed by the application of artificial tears and eye lubricants. In severe cases, surgical correction may be required. Administration of retinoids has a keratolytic effect, helps in the elimination of scales, and prevents hyperkeratosis of the skin. Isotretinoin and retinoids have both proven to be effective in these cases. The prophylactic use of antibiotics is not recommended. Ophthalmological management of ectropion is important for the prevention of conjunctivitis and keratitis. Pain management should be taken care of. A water bed is useful, and the prophylactic use of mild analgesics before daily bathing and skin care is appropriate. Sedation with opioids may be indicated when handling the child is very painful. The use of retinoids may be considered when the shedding of the collodion membrane is much delayed (after 3 weeks) because of the infectious risk associated with the humidified milieu in the incubator and mostly because of the intrinsic severity of the underlying ichthyosis. In that case, acitretin may be given at 0.5–0.75 mg/kg per day. The newborn was also started on L-thyroxine at a dose of 10 micrograms per kg per day, and later the dose was titrated. The long-term prognosis is difficult to address at

birth, and the compromised skin status of the child is difficult to accept for the family.

Figure 2: Collodion baby with hypothyroidism with improving scaly condition.

DISCUSSION

The term "collodion baby", also known sometimes as "collodion foetus", was first used in 1884 to describe the physical appearance of a newborn with a parchment-like membrane covering the whole body.[1] It is a very rare condition, occurring in 1 in 50000 to 1 in 100000 live births.[2] The collodion babies are wrapped in an oiled parchment-like membrane that resembles cellophane and covers the entire body. Affected neonates have eversion of the eyelid (ectropion), slightly everted lips, restricted extremity and digit movements due to pseudo-contractures, a lack of eyebrows, sparse hairs on the head, and a deformed nose and ears due to nasal and ear cartilage hypoplasia. The babies had poor sucking, distal limb ischemia, and extremity oedema, along with flattening of the ears and nose, as well as fixation of the lips in an O shape. Later, the membrane cracks and begins to desquamate. The collodion membrane sheds in the weeks following birth, exposing the underlying skin disorder.

In the long run, about 75% of collodion baby cases will develop autosomal recessive congenital ichthyosis (lamellar ichthyosis or congenital ichthyosiform erythroderma).[3] In only 10% of these cases, the membrane sheds and the underlying skin remains normal for the rest of the patient's life.[3] In the remaining 15% of cases, associations with various entities such as ichthyosis vulgaris, trichothidystrophy, and metabolic and endocrine disorders involving keratinization disorders are observed.[4]

Lamellar ichthyosis (LI) is a type of congenital ichthyosis that is characterised by hyperkeratosis. The incidence of LI is one per 140,000–300,000 people. LI cases are frequently caused by genetic mutations such as transglutaminase 1 (TGM1), NIPAL4, ALOX12B, ALOXE3, ABCA12, CYP4F22, PNPLA1 (OMIM 615024), and CERS3 (OMIM 615023).[5] TGM1 has 15 exons and is found on chromosome 14q11.2 (GenbankNM-000359.22).[6] The TGM1 gene mutation is located on chromosome 14q12 and is the most common cause of collodion babies.[7] Because autosomal recessive congenital ichthyosis is the most common presentation in collodion babies, genetic counselling and prenatal diagnosis are required in affected families. The earliest diagnosis is possible between the 10th and 12th weeks of gestation using genomic PCR on chorionic villous samples or between the 15th and 18th weeks using amniocentesis.[8] A foetal skin biopsy can be performed between the 18th and 20th weeks of pregnancy.[9] Prenatal diagnoses by ultrasound (2D and 3D) imaging can detect features suggestive of a harlequin foetus in the 1st to 2nd trimester of pregnancy. Ultrastructural abnormalities can also be detected using a foetal skin biopsy.[10] A routine skin biopsy is not advised.[6] If molecular testing is performed, the TGM1 gene should be examined first. If this is negative, gene analysis for ALOX12B, ALOXE3, and NIPAL4 can be performed.[11]

CONCLUSION

Collodion baby is a rare congenital phenotypic disorder whose prognosis depends on the initial presentation and timely neonatal management. To prevent long term morbidity and mortality, there is a need for early diagnosis and a multi-disciplinary approach. The patients should be counselled about the risk of recurrence in the subsequent pregnancies based on the genetic counselling of the families with children affected by congenital ichthyosis and made aware of the possible strategy for undergoing prenatal diagnosis in the subsequent pregnancies. The Collodion baby with hypothyroidism and its timely management of the associated abnormalities also play an important role in the baby's prognosis.

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REFERENCES

1. Herman ML, Farasat S, Steinbach PJ, Wei MH, Toure O, Fleckman P, et al. Transglutaminase-1 (TGM1) Gene Mutations in Autosomal Recessive Congenital Ichthyosis: Summary of Mutations (Including 23 Novel) and Modeling of TGase-1. Hum Mutat. 2009;30(4):537-47.
2. Dyer JA, Spraker M, Williams M. Care of the newborn with ichthyosis. Dermatol Ther. 2013;26(1):1-15.
3. Van Gysel D, Lijnen RLP, Moekti SS, de Laat PCJ, Oranje AP. Collodion baby: a follow-up study of 17 cases. J Eur Acad Dermatol Venereol JEADV. 2002;16(5):472-5.

4. Dyall-Smith D, Marks R. Dermatology at the Millennium. Taylor & Francis. 1999;844.
5. Fachal L, Rodríguez-Pazos L, Ginarte M, Carracedo A, Toribio J, Vega A. Identification of a novel PNPLA1 mutation in a Spanish family with autosomal recessive congenital ichthyosis. Br J Dermatol. 2014;170(4):980-2.
6. Fachal L, Rodríguez-Pazos L, Ginarte M, Beiras A, Suárez-Peñaranda JM, Toribio J, et al. Characterization of TGM1 c.984+1G>A mutation identified in a homozygous carrier of lamellar ichthyosis. Int J Dermatol. 2012;51(4):427-30.
7. Sharma D, Gupta B, Shastri S, Pandita A, Pawar S. Collodion Baby with TGM1 gene mutation. Int Med Case Rep J. 2015;8:205-8.
8. Akiyama M, Titeux M, Sakai K, McMillan JR, Tonasso L, Calvas P, et al. DNA-Based Prenatal Diagnosis of Harlequin Ichthyosis and Characterization of ABCA12 Mutation Consequences. J Invest Dermatol. 2007;127(3):568-73.
9. Rimoin L, Graham JM. Ichthyotic skin disorders in the neonate. Clin Pediatr (Phila). 2012;51(8):796-800.
10. Rossi G, Mesia D. Management of harlequin ichthyosis in low-income countries. Ann Trop Paediatr. 2011;31(3):247-9.
11. Vahlquist A, Bygum A, Gånemo A, Virtanen M, Hellström-Pigg M, Strauss G, et al. Genotypic and clinical spectrum of self-improving collodion ichthyosis: ALOX12B, ALOXE3, and TGM1 mutations in Scandinavian patients. J Invest Dermatol. 2010;130(2):438-43.

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