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CONGENITAL LAMELLAR ICHTHYOSIS IN NEWBORN: A CASE REPORT

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ABSTRACT: The term Congenital Lamellar Ichthyosis is used for newborns in whom all the body surface is covered by thick skin sheets, so called "collodion membrane". Ichthyosis is an infrequent clinical entity worldwide (1:300,000 births). The collodion membrane is the result of an epidermal developmental dysfunction. The collodion membrane is composed of thick skin sheets which resemble translucent, tight parchment paper. In almost all of the collodion membrane cases an autosomal recessive ichthyosiform disease is implicated. Especially, in cases of lamellar ichthyosis, congenital ichthyosiform erythroderma and harlequin ichthyosis frequent association with collodion baby formation has been well documented. Clinically, the collodion babies may encounter dehydration, electrolyte imbalance, temperature imbalance and increases risk of sepsis because of relatively severe skin damage. Therefore, morbidity and mortality rates are fairly high in these cases. Conclusively, these newborns should be monitored carefully in intensive care units and is difficult to diagnose in antenatal period.

KEYWORDS: Congenital Lamellar Ichthyosis, Collodion baby, Ectropion.

INTRODUCTION: The term was first introduced by Hallopeau and Watelet.¹ The first clinical description of collodion membrane by Pérez in 1880 continues to be valid. "The baby's skin is replaced by a cornified substance of uniform texture, which gives the body a varnished appearance".² As the name suggests, the term "collodion baby" refers to a phenotype that can be characterized by a yellow, shiny, tight parchment-like membrane stretched over the skin. Observers may sometimes use the descriptor "dipped in hot wax."³ The most important clinical data concerning collodion baby is the presence of disseminated or generalized 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.⁴

Harlequin ichthyosis or harlequin fetus, is a severe generalized disorder of epidermal differentiation characterized by massive generalized hyperkeratosis and usually death in the perinatal period. Ultrastructurally, harlequin ichthyosis has an absence of normal lamellar bodies. At birth, the thick stratum corneum results in constrictive deformation, with severe ectropion and eclabium. Vital functions such as respiration and swallowing are compromised. Survival in the neonatal period is rare. Patients with harlequin ichthyosis represent the most severe end of the phenotypic spectrum of autosomal recessive ichthyosis.⁵

The clinical types of ichthyosis depend on the mode of inheritance as well as clinical and anatomic/pathological data.¹ There are several subtypes of each group. Among the true ichthyoses are three subgroups as follows: 1) Autosomal dominant ichthyosis (ichthyosis vulgaris,

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ichthyosis simplex) 2) X-linked recessive ichthyosis (ichthyosis nigricans, ichthyosis of the male, saurodermia) and 3) Autosomal recessive ichthyosis (lamellar ichthyosis, non-bullous congenital ichthyosiform erythroderma).⁶ Rarely there may be an association with bullous congenital ichthyosiform erythroderma, Gaucher's disease and Sjögren-Larsson syndrome. Furthermore, a new form of the disease with an autosomal recessive inheritance called "self-healing collodion baby" has been notified where the newborn completely recovers in a couple of weeks.^{3, 7}

CASE REPORT: A term male baby was delivered by LSCS and cried immediately after birth with birth weight 2.63kg, length 45cms, head circumference 34 cms. No resuscitation was required. Admitted in NICU in view of abnormal looking skin and was started orogastric feeds and antibiotics. History of similar complaint was present in the second sibling. This was the third sibling, first sibling had no similar complaints. There was history of consanguineous marriage with positive family history and being healthy parents.

On clinical examination baby was presented with a thin collodion membrane that was found over the majority of the skin surface. Baby was active and alert, vitals stable. On physical examination there was patchy areas of erythema with large adherent brown scales over the body. Fissures were present in flexoral areas and periorbital swelling was present since birth.

Strict temperature control was maintained. Sepsis screen sent for the baby was negative. Biochemical parameters like liver function tests, renal function tests and serum calcium levels were within normal range.

The membrane started peeling from day 2 itself, revealing red colored skin underneath. Baby received breastfeeding from day 3 of life. Strict asepsis was maintained whenever dealing with the baby and antibiotics were given for 3 days.



COLLODION BABY AT BIRTH

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COLLODION BABY ON FOLLOWUP

COLLODION BABY: Konaseema Institute of Medical Sciences, Amalapuram, AP, India.

DISCUSSION: The term collodion baby applies to newborns that appear to have an extra layer of skin (known as a collodion membrane) that has a collodion-like quality. It is a descriptive term, not a specific diagnosis or disorder (as such, it is a syndrome).⁸ This is exactly noted in our patient who had extra layers of transparent skin.

The natural course of collodion membrane is intriguing. For instance, approximately 75% of collodion baby will go on to develop a type of autosomal recessive congenital ichthyosis, either lamellar ichthyosis or congenital ichthyosiform erythroderma).⁹ Another 10% of cases the baby sheds this layer of skin and has normal skin for the rest of its life.¹⁰ This is known as self-healing collodion baby. This is akin to our series. The remaining 15% of cases could stem from variety of diseases involving keratinization disorders.⁹

The major cause of collodion baby syndrome is not well known. However it has been known to be inherited in an autosomal recessive fashion.¹¹ Placental insufficiency and post maturity have also been implicated in some forms of collodion membrane formation. This could be due to the effects of DNA repair and transcription gene abnormalities in human pre-natal life. Trichothiodystrophy (TTD), a rare (affected frequency of 10–6) recessive disorder caused by mutations in genes involved in nucleotide excision repair (NER) pathway has also been implicated.

Due to the impairment of the skin barrier function, collodion babies are at risk for a number of complications including dehydration, hypothermia, skin infections, fissures, conjunctivitis, sepsis, hypernatremic dehydration, and constrictive bands of the extremities resulting in vascular compromise and edema.^{1, 12} The edema in the patient described here was thought to be due to either hypoproteinemia or mechanical compression by the collodion membrane. One study showed that the transepidermal water loss in collodion babies can be six to seven times higher than that through normal skin.¹³ Skin impairment gives rise to transepidermal water loss (TEWL), percutaneous infection, and toxicity. Accurate intake and output monitoring is essential, as is the accurate monitoring of electrolytes for hypernatremia and the appropriate adjustments of fluid intake. Therefore, it is essential that collodion babies be

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placed in a humidified incubator soon after birth to prevent hypernatremic dehydration and hypothermia. The patient described here was managed accordingly with the use of emollients and prophylactic antibiotics.

CONCLUSION: Because this is a rare disease it is indispensable to have very clear and precise information on the steps to follow and the complications that may arise. In order to determine the etiologic cause for the collodion membrane, a protocol must be established so that appropriate measures can be taken months or years following the shedding of the collodion membrane.

An incubator provides a humidified, neutral temperature environment. Other supportive treatments such as intravenous fluid and tube feeding are often necessary. The aim is to keep the skin soft and attempt to reduce scaling. The collodion membrane should not be debrided (pulled off).

In our experience through this case study, we suggest careful attention to skin care, minimal use of skin care products, and meticulous attention to preventing infection when caring for a newborn with collodion baby syndrome.

REFERENCES:

1. D. van Gysel, R. L. P. Lijnen, S. S. Moekti, P. C. J. De Laat, and A. P. Oranje, "Collodion baby: a follow-up study of 17 cases," *Journal of the European Academy of Dermatology and Venereology*, vol. 16, no. 5, pp. 472–475, 2002.
2. Cortina WJ, Cruz MJ, Villalobos OA, Espinoza MA. Ictiosis congenital (feto arlequín). *Bol Med Hosp Infant Mex* 1975; 32: 699-702
3. M. R. Judge, "Collodion baby and Harlequin ichthyosis," in *Textbook of Pediatric Dermatology*, J. Harper, A. Oranje, and N. Prose, Eds., pp. 118–125, Blackwell, Malden, Mass, USA, 2nd edition, 2006.
4. O'Connell JP. A collodion baby. *Proc R Soc Med* 1977; 70: 212-13
5. Burdette-Taylor S. Case study: harlequin fetus, a disease of cornification. *Ostomy Wound Manage* 1994; 40: 14 –9.
6. Arenas R. Dermatología. Atlas, Diagnóstico y Tratamiento. México: McGraw-Hill-Interamericana; 1996. p. 225-30.
7. Shwayder T, Akland T. Neonatal skin barrier: structure, function and disorders. *Dermatol Therapy* 2005; 18: 87-103. PMID: 15953139.
8. Larrègue M, Ottavy N, Bressieux JM, Lorette J. "[Collodion baby: 32 new case reports]" (in French). *Ann Dermatol Venereol* 1986; 113: 773–85.
9. *Dermatology at the Millennium*. By Delwyn Dyall-Smith, Robin Marks, Page 586, Published by Informa Health Care, 1999, ISBN.
10. Van Gysel D, Lijnen RL, Moekti SS, de Laat PC, Oranje AP. "Collodion baby: a follow-up Study of 17 cases". *J Eur Acad Dermatol Venereol* 2002; 16: 472–5.
11. Moslehi R, Signore C, Tamura D, Mill JL, DiGiovanna JJ, Tucker MA et al. Adverse effects of trichothiodystrophy DNA repair and transcription gene disorder on human fetal development. *Clin Genet* 2010; 77: 365–373.

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12. J. B. Roberts and D. Adelson, "Case report: prolonged collodion membrane causing constrictive bands of the digits and treatment," *Dermatology Online Journal*, vol. 16, no. 1, article 15, 2010.
13. L. Buyse, C. Graves, R. Marks, K. Wijeyesekera, M. Alfaham, and A. Y. Finlay, "Collodion baby dehydration: the danger of high transepidermal water loss," *British Journal of Dermatology*, vol. 129, no. 1, pp. 86–88, 1993.

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