

Collodion baby: Clinical spectrum and outcome in a series of 5 cases

Archana Kashyap¹, Bhavna Anand¹, Ininaki Lyngdoh²

¹Associate Professor, Department of Pediatrics VMMC & Safdarjung Hospital, New Delhi, India. ²Senior Resident, Department of Pediatrics VMMC & Safdarjung Hospital, New Delhi, India

Abstract

Collodion baby is a rare congenital disorder of cornification characterized by a taut, parchment-like membrane covering the newborn's body, with an estimated incidence of 1 in 50,000–100,000 births. Most cases follow an autosomal recessive inheritance pattern. This study included five patients with clinical suspicion of collodion disease who presented over a 10-month period. Clinical features, laboratory investigations, treatment, complications, and outcomes were evaluated. Baseline investigations were performed in all patients, while genetic testing was planned whenever feasible; however, financial constraints limited genetic evaluation. All patients were male and presented with features of congenital ichthyosis, frequently accompanied by complications requiring hospital management. Patients presenting during the neonatal period had higher mortality, with sepsis being the predominant cause of death. Early clinical recognition and multidisciplinary involvement facilitated diagnosis and guided appropriate supportive management. Prompt recognition and management of complications are essential for improving outcomes in patients with collodion baby.

Keywords: Collodion, genetic testing, ichthyosis, incidence, inheritance pattern, sepsis.

Received: 17 September 2026

Revised: 22 September 2026

Accepted: 28 September 2026

Published: 10 October 2026

INTRODUCTION

Collodion baby is a rare congenital disorder whereby patients are brought with presenting features of a taut-like membrane covering the whole body, which occurs secondary to a disorder of cornification.^[1]

It is characterized by hyperkeratosis, severe erythroderma, and a slight desquamation pattern.^[2] The incidence of collodion baby is estimated to be 1 in every 50,000–100,000 births.^[3]

The exact cause is unknown, but in most cases, it was found to be an autosomal recessive disorder. Autosomal recessive congenital ichthyoses encompass several forms of non-syndromic ichthyosis, which can vary in presentation and severity of the disease. The most severe forms, occasionally fatal, are harlequin ichthyosis, lamellar ichthyosis, and congenital ichthyosiform erythroderma.^[4] Lamellar ichthyosis is an AR type of congenital ichthyosis characterized by hyperkeratosis. The TGM1 gene was associated with the self-improving type of collodion ichthyosis, with 70–90% of individuals presenting with lamellar ichthyosis.^[5]

All cases of Collodion Disease who presented to the Paediatric Outpatient Department and Emergency Department of VMMC and Safdarjung Hospital between October 2023 and December 2023 were enrolled in the study [Table 1]. History, examination, and baseline investigations were performed for all patients who were enrolled in the study. All patients were planned for genetic testing when feasible; however, due to financial constraints, genetic testing was not possible for all the patients.

Case 1: A patient at 8 hours of life with generalized cracked skin, including ectropion. Born of vaginal delivery, the baby weighed 2.684 kg at 38 weeks with 35 cm of head circumference and 52 cm length. There was a history of a similar disease in a previous sibling who expired on day 4 of life. There was history of consanguinity. On examination, the patient had respiratory distress with an initial diagnosis of transient tachypnea of the newborn. On general physical examination, the patient had thick parchment-like skin with peeling of the skin in the extremities along with fissures. Ectropion and eclabium were also noted in the patient. The patient had thick-like secretions in the external auditory canal. A sepsis screen was sent at the time of admission of the patient and was negative. CRP had come out negative, and the complete blood count was initially normal, but a repeat sample showed a sepsis score of more than or equal to 3, while blood and pus cultures were both negative. Sepsis was attributed to late-onset neonatal sepsis, for which the patient had received antibiotics. Genetic testing for this patient was not feasible due to financial constraints, as the testing was not available within the hospital. The child was discharged. [Figure 1]

Address for correspondence: Dr. Archana Kashyap
Associate Professor Department of Pediatrics VMMC & Safdarjung Hospital, New Delhi, India
E-mail: Archana425@gmail.com

DOI:
10.21276/amt.2026.v13.i3.1114

How to cite this article: Kashyap A, Anand B, Lyngdoh I. Collodion baby: Clinical spectrum and outcome in a series of 5 cases. *Acta Med Int.* 2026;13(3):603-607.



Figure 1: Case 1



Figure 2: (a) and (b): Case 2

Case 2: A seven-year-old male child, first-born male son of a family with three other female siblings, presented to our Emergency Department with complaints of abdominal pain for 4 to 5 weeks with multiple episodes of passage of black tarry stools. The patient had abdominal pain, which was dull in nature, with multiple episodes of black tarry stools. On examination in the Emergency department, the patient's vitals were taken, and the patient was found to be hemodynamically stable. On further examination, the patient had coarse facies with thick skin associated with peeling of skin at the joints [Figure 2a, 2b]. The patient also had thick-like secretions in the external auditory canal. The height of the patient was 138 cm and the weight of the patient was 25 kg, which was normal for age. On systemic examination, no abnormality was detected. Initial baseline investigations taken at the time of admission were found to be normal, with a normal coagulation profile. Gastric aspirate for tuberculosis was also sent, which came out negative. Genetic testing was done, which revealed a TGM1 gene mutation of autosomal recessive type of inheritance with a homozygous genotype. This was consistent with the suspected diagnosis of congenital ichthyosis, an autosomal recessive disease. Gastrointestinal endoscopy was performed for melena and revealed that the child had multiple ulcers with irregular, punched-out margins in the

duodenum. The histopathology report was awaited but the child got discharged against medical advice.

Case 3: A 5-day-old male neonate presented with respiratory distress for 2 days, lethargy for 1 day, and generalized scaly lesions since birth. There was no similar family history or consanguinity; a previous pregnancy had ended in spontaneous abortion at 2 months.

On examination, the neonate was in shock, with delayed capillary refill and poor peripheral pulses. General examination revealed generalized scaling with thick secretions around the eyes and ears. Bilateral air entry was present with crepitations, and the patient was lethargic. The sepsis screen was positive, and blood culture grew *Staphylococcus aureus*, which was sensitive to all tested antibiotics. Serum sodium was 156 mmol/L, potassium 4.2 mmol/L, blood urea nitrogen 53 mg/dL, and creatinine 0.3 mg/dL. Cerebrospinal fluid analysis was suggestive of meningitis, with elevated cell count and protein levels.

Initial management included intravenous access, normal saline resuscitation, and empirical antibiotics, which were subsequently escalated following the diagnosis of meningitis. The blood culture-confirmed organism was *Staphylococcus aureus*. Genetic testing was planned but could not be performed due to financial constraints. The neonate expired on day 7 of hospital admission. [Figure 3]



Figure 3: Case 3



Figure 4(a): Case 4



Figure 4(b): Case 4

Case 4: A male child was brought on day 9 of life following referral to our hospital in view of scaly lesions all over the body, ectropion, and eclabium since birth. There was no history of consanguinity or similar illness in the family. On examination, the patient was found to be hemodynamically stable. General physical examination revealed scaly lesions over the body along with ectropion and eclabium. The sepsis screen was positive, and the possibility of sepsis was considered. A blood culture report was also sent, which showed no growth. Lumbar puncture showed features of meningitis; however, no growth was noted in the CSF. The patient received multidisciplinary referrals and treatment from dermatology, ophthalmology, and plastic surgery for the various complications. Dermatology advised treatment with liquid paraffin twice a day. Ophthalmology examination revealed bilateral keratosis; strict padding and patching along with eye drops

were advised for the patient. Whole-exome sequencing for the patient has been sent. [Figure 4a and 4b]

Case 5: A 10-month-old male child, born to a primigravida, presented to our OPD with complaints of diffuse scaly lesions all over the body since birth. The patient also came with complaints of thick secretions from the eyes and aural mucosa. The patient had these complaints since 15 days of birth, followed by multiple visits to a local practitioner, for which the patient received ointment for local application. There was no history of consanguinity, similar family history, or sibling death.



Case 5: Figure 5a, 5b

On examination, the patient was hemodynamically stable. Eye examination revealed ectropion over the right eye with thick mucoid secretions over bilateral auditory canals. Systemic examination was within normal limits. The patient was given multidisciplinary care, including skin care with petroleum jelly treatment. For this patient, isotretinoin creams were planned. An eye referral was also sent for ectropion, along with an otolaryngologist referral for mucoid secretions in the ear. The patient was planned for genetic testing. However, due to financial constraints, genetic testing was not done in this patient. He was discharged and was lost to follow-up in the subsequent OPD visits. [Figure 5a, 5b]

Table 1: Case details

	CASE 1	CASE 2	CASE 3	CASE 4	CASE 5
Age of presentation	8hours of life	7years	Day 5 of life	Day 9 of life	10months
Gender	Male	Male	Male	Male	Male
Consanguinity	yes	No	no	no	no
Chief presentation:					
Scaly lesion	Yes	Yes	Yes	Yes	Yes
Ectropion	No	No	Yes	Yes	No
Eclabium	No	No	No	Yes	No
Fever	yes	No	yes	yes	No
Investigation					
TLC	9200cells/L	7000cells/L	3200cells/L	5600cells/L	8960cells/L
CRP	Negative	negative	Positive	Negative	Negative
Blood c/s	No growth	No growth	Staph aureus	No growth	No growth
Genetics	Not done	TGM1 gene	Not done	Awaited	Awaited
Outcome	Discharged	Discharged	Death	Admitted	Discharged

DISCUSSION

We present here five cases of collodion babies who presented at different time periods of their life, ranging from as early as 8 hours of life up to 7 years.

The reported age at presentation in previous studies has generally been within the neonatal period. Bouab et al,^[1] reported a premature infant born at 8 months (35 weeks) gestation who presented at birth. Guo et al,^[6] described a full-term infant (38+3 weeks) who presented 30 minutes after birth with a tense collodion membrane. Quazi et al,^[2] reported a full-term newborn (38 weeks gestation) who

presented at birth with a glossy collodion membrane. Amaresh et al,^[4] described an infant (37+5 weeks) who presented immediately at birth and was subsequently followed until 4 months of age. Similarly, Wang L et al,^[5] reported a premature infant (35+6 weeks) who presented within 10 minutes of birth with skin cracking, diffuse erythema, and skin tightness.

The presentation of collodion babies is typically immediate, occurring at birth or within the first few minutes to 30 minutes of life. This reflects its congenital origin, with abnormal epidermal barrier formation occurring in utero. The characteristic tight, shiny, parchment-like membrane is

therefore evident at delivery, while respiratory, feeding, fluid-loss, and infection-related complications may necessitate early neonatal care.^[7]

All the present cases were males. In comparison, Bouab et al,^[1] reported a female infant, Guo et al,^[6] reported a female infant, and Quazi et al,^[2] reported a female newborn, whereas Amaresh et al,^[4] and Wang L et al,^[5] reported male infants. Thus, the present case series showed an exclusively male presentation, whereas the previously reported cases included both male and female infants.

Collodion babies show no established sex predilection because the condition is predominantly associated with autosomal recessive mutations, including TGM1, ALOX12B, ALOXE3, and CERS3. Because the responsible genes are non-sex-linked, there is no genetic predilection for either sex, and males and females are affected with equal probability.^[8]

Consanguinity remains a concern in such congenital disorders. In our present case series a single case was born out of consanguinous marriages, while other cases had no consanguinity in the marriages.

However, Bouab et al,^[1] Guo et al,^[6] Quazi et al,^[2] Amaresh et al,^[4] and Wang L et al,^[5] reported non-consanguineous parental unions in their respective cases. Thus, like most cases in the present series, consanguinity was not reported in any of these previously published cases.

Parental consanguinity represents a well-established vulnerability for autosomal recessive dermatological conditions, including collodion baby syndrome, by elevating the probability that offspring inherit matching homozygous genetic variants. Nonetheless, affected neonates can equally manifest in unrelated couples through compound heterozygous mutation.^[6]

Genetic analysis remains important whereby the TGM gene was mutated in one case, while others did not undergo genetic analysis.

In comparison, Guo et al,^[6] performed whole-exome sequencing followed by Sanger sequencing, which identified compound heterozygous mutations in TGM1. Similarly, Wang L et al,^[5] performed clinical exome sequencing and identified compound heterozygous TGM1 mutations. Amaresh et al,^[4] performed next-generation sequencing, which identified a homozygous single-base-pair deletion in exon 9 of the CERS3 gene. Thus, genetic evaluation in previously reported cases identified pathogenic variants involving TGM1 and CERS3, whereas genetic testing was performed in only one case in our series. Genetic analysis helps establish the underlying etiology of the collodion baby phenotype and differentiate ARCI subtypes when clinical features overlap. It can provide prognostic information regarding disease evolution, facilitate genetic counseling and assessment of recurrence risk, identify novel pathogenic variants, expand the mutational spectrum, and potentially support future development of targeted therapies.^[4]

The chief presentations comprised fever and scaly lesions. As for outcomes a single case of death was reported. Blood culture showed *Staphylococcus aureus* and the patient had meningitis. In comparison, Bouab et al,^[1] found a thick,

parchment-resembling membrane with the neonate succumbing within 10 hours of life. Guo et al,^[6] reported generalized edema, and tense, transparent membrane presenting at birth; with no pathogenic growth. Likewise, Quazi et al,^[2] observed a widespread, reflective, translucent parchment membrane marked by fissuring and peeling, with uneventful discharge. Amaresh et al,^[4] noted erythematous, reflective, tense, parchment-like cutaneous features alongside breathing difficulty; subsequent blood cultures isolated *Pseudomonas* and *Klebsiella*, prompting intravenous antibacterial therapy, treated and followed by healthy discharge on day 21. The outcome remained varied among cases with early diagnosis and infection needing IV antibiotics.

Limitations: Genetic evaluation could not be done in all patients because of financial constraints, restricting molecular characterization and genotype–phenotype correlation. The cases were observed over a limited period, and long-term follow-up was unavailable.

CONCLUSION

Collodion baby is a rare congenital disorder with variable age at presentation and clinical severity. In our five-case series, presentations ranged from 8 hours of life to 7 years, highlighting that affected children may present beyond the immediate neonatal period. All cases were male, although the condition has no established sex predilection. One patient had confirmed TGM1 mutation, while genetic testing was unavailable or pending in the remaining cases. Neonatal complications, particularly infection and sepsis, contributed to adverse outcomes. Early recognition, appropriate supportive care, multidisciplinary management, and genetic evaluation when feasible are significant in establishing diagnosis and individualized management.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

REFERENCES

1. Bouab M, Wajih O, Assal A, Jalal M, Lamrissi A, Bouhya S. Collodion baby: A rare case report. *Int J Surg Case Rep*. 2023;112:108930.
2. Quazi S, Singh A, Khan K, Biyani U. A case report of a collodion baby: an autosomal recessive genodermatosis. *Cureus*. 2023;15(4):e37418.
3. Ana EG, María MG, Alejandro JD, Laura SF. Self-healing collodion baby: a rare condition. *Rev Pediatr Aten Primaria*. 2025;27(105):77-9.
4. Amaresh P, Teotia S, Garg AK, Kondaparthi N. From phenotype to genotype: a collodion baby case linked to CERS3 mutation. *J Neonatal*. 2025;39(2):196-9.
5. Wang L, Li X, Yang L, Xi H, Li L. A novel mutation in the transglutaminase-1 gene identified in a collodion baby: A case report. *J Int Med Res*. 2025;53(10):1-11.
6. Guo Y, Xiao Z, Hu X, Liu Y, Chen G. A case report and literature review of self-improving collodion baby in the newborn. *Medicine (Baltimore)*. 2025;104(14):e42045.

7. Stolarczyk A, Bender A, Plovanich M, El-Feghaly J. Acute management of constriction bands in a newborn with collodion membrane with early manual release. JAAD Case Rep 2023;38:144-7.
8. Zdraveska N, Kostovski A, Sofijanov A, Jancevska S, Damevska K. Collodion phenotype remains a challenge for neonatologists: A rare case of self-healing collodion baby. Clin Case Rep 2022;10(7):e6158.