# Unusual presentation of congenital chloride diarrhea mimicking colovesical fistula: a case report and literature review

Mohammed AlMesaibli[1*], Tuqa A. AlSinan[2], Adeeba Sajid[3], Madiha Jamal[3], Tehreemah Raziq[3]

**European Journal of Medical Case Reports**
Volume 8(2):41–43
DOI: 10.24911/ejmcr.173-1711931032



## ABSTRACT

**Background:** Congenital chloride diarrhea (CCD) is a rare autosomal recessive condition characterized by lifelong watery diarrhea. CCD is challenging to diagnose because of its severe clinical presentations and differentials. Therefore, it is crucial to be diagnosed early and receive appropriate management as it causes early death if left untreated or due to complications.

**Case Report:** A Saudi newborn baby boy was passing a large amount of urine through the anal canal and was otherwise healthy. He was referred to our center with a high suspicion of a colovesical fistula. Diagnosed with CCD by the exclusion of fistula after confirmation with contrast study.

**Conclusion:** This case highlights an unusual presentation of CCD mimicking a colovesical fistula. To the best of our knowledge, no similar case has been reported in the literature, and we believe this is the first Saudi case of CCD mimicking a colovesical fistula. It underscored the diagnostic challenges of this rare disorder and emphasized the importance of early recognition and accurate diagnosis to prevent such complications.

**Keywords:** Congenital chloride diarrhea, colovesical fistula, rare genetic, autosomal recessive, Pediatrics.

**Received:** 01 April 2024 **Accepted:** 13 April 2024 **Type of Article:** CASE REPORT **Specialty:** Pediatrics

**Correspondence to:** Mohammed AlMesaibi
*Section of Pediatric Surgery, Department of Surgery, King Saud University Medical City, Riyadh, Saudi Arabia.
**Email:** medosurg@gmail.com
*Full list of author information is available at the end of the article.*

## Background

Congenital chloride diarrhea (CCD) is a rare autosomal recessive condition characterized by lifelong watery diarrhea. It is caused by mutations in the SLC26A3 gene on chromosome 7q31, which encodes for Cl-/HCO3 exchanger, leading to its absence and thus Cl-rich watery diarrhea [1,2]. The incidence of CCD has been documented in a range of ethnic populations, such as Finland, Poland, and Saudi Arabia, carrying the same mutation due to the genetic founder effect. Saudi Arabia has a prevalence estimated at around 1:5500, though it may be considerably higher [2,3]. Patients usually present with polyhydramnios and intestinal dilatation, which is detected prenatally, requiring hospitalization and rigorous hydration due to potential complications. Shortly after delivery, patients frequently experience severe diarrhea, which can lead to dehydration and electrolyte imbalances [1,4].

The condition manifests itself by causing hyponatremia, hypokalemia, hypochloremia, and metabolic alkalosis [4]. The patient is treated with lifelong salt substitution treatment to prevent dehydration as well as allow average growth and development as the intestinal defect cannot be fixed. In most situations, additional therapies such as butyrate and cholestyramine successfully reduce diarrhea [1-3]. CCD is challenging to diagnose because of its severe clinical presentations and differentials. Therefore, it is crucial to be diagnosed early and receive appropriate management [1]. CCD causes early death if left untreated or due to complications such as impaired renal function, nephrocalcinosis, and end-stage renal disease [1-3]. This case report represents a case of a Saudi newborn boy with CCD and his management.

## Case Presentation

A Saudi newborn baby boy who was born in August 2021 with a condition of passing a large amount of urine through the anal canal and was otherwise healthy [Video 1] (Hyperlink: https://youtu.be/uVjN39uTCKw). The baby was born via spontaneous vaginal delivery, full-term, with an average birth weight of 3.1 Kg, passed meconium within the first 24 hours, and uneventful antenatal history. The parents are cousins, and the mother is 30 years old and healthy, with no known medical or genetic illnesses. There is a family history of similar presentation of two siblings at birth who unfortunately did not survive.

Initially, he was admitted to a local hospital with high suspicion of a colovesical fistula, which prompted close monitoring in the neonatal intensive care unit and specialized care to address dehydration and stabilize the infant. The baby was referred to a tertiary center for further pediatric surgical consultation and management. Further investigations and physical examination yielded inconclusive results.

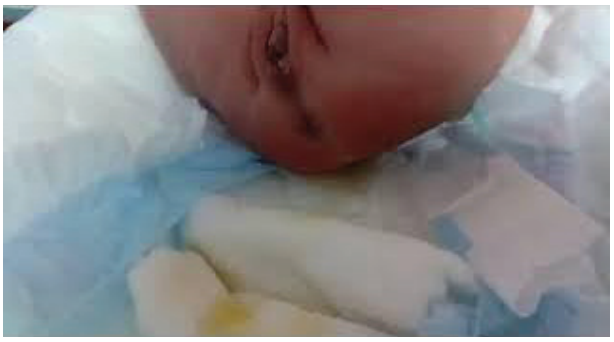
After excluding colovesical fistula and bowel obstruction, blood laboratory tests revealed hyponatremia (124mml/l), hypokalemia (2.5mEq/l), and a pH level was 7.25. Urine analysis was unremarkable. However, stool analysis revealed a high elevation of chloride level (95mmol/l), which confirmed the diagnosis of CCD. The colovesical fistula was excluded and confirmed by the absence of a fistula based on a contrast study. The infant's management included resuscitation and medical intervention without the need for surgical intervention. The medical team closely monitored the newborn's hydration status and implemented appropriate electrolyte replacement therapy. The patient responded well to treatment, and the prognosis was favorable.

## Discussion

CCD is a rare autosomal recessive disorder characterized by chronic diarrhea due to impaired chloride absorption in the intestines. Diagnosis can be challenging, but early recognition is crucial for effective management and prevention of complications. The reviewed literature sheds light on various aspects of CCD, emphasizing the importance of early diagnosis and management. The literature highlights the significance of a high level of suspicion for CCD in infants presenting with chronic diarrhea, particularly in regions with a high prevalence of consanguineous marriages. Antenatal ultrasound findings, such as dilated bowel loops, can serve as an important diagnostic clue for CCD. This early identification allows for prompt investigations and the initiation of appropriate management strategies to prevent complications [4-7].

A case report and literature review underscored the diagnostic challenges of CCD and its overlaps with other conditions, such as Bartter syndrome and ulcerative colitis. This emphasizes the need for a comprehensive evaluation, including genetic testing, to confirm the diagnosis of CCD and avoid misdiagnosis. Early recognition is vital for preventing serious complications [5]. In addition, the co-occurrence of CCD with other rare genetic disorders, such as pendred syndrome, highlights the complex genetic nature of these conditions. The presence of multiple rare recessive traits in consanguineous couples can increase the risk of CCD in their progeny. Understanding these genetic associations can aid in accurate diagnosis and counseling [8].

The review of CCD's clinical features and management strategies emphasizes the importance of a multidisciplinary approach and close follow-up. CCD is a multisystemic disorder with heterogeneous complications requiring individualized management. Optimal therapeutic strategies and regular monitoring are essential for improving outcomes and maintaining a good quality of life [1,6-9].



***Video 1.*** *The presentation of the infant passing urine through the anal canal.*

Available from: https://youtu.be/uVjN39uTCKw

## Conclusion

This case report highlights an unusual presentation of CCD mimicking a colovesical fistula. To the best of our knowledge, no similar case has been reported in the literature, and we believe this is the first Saudi case report with an unusual presentation. It underscored the diagnostic challenges of this rare disorder and emphasized the importance of early recognition and accurate diagnosis to prevent such complications. A multidisciplinary approach and individualized management strategies are essential for optimizing outcomes in CCD patients. Further research and awareness are needed to improve understanding and facilitate timely intervention in atypical CCD presentations.

### What is new?

This case shows the unusual presentation of passing urine through the anus with high suspicion of colovesical fistula. The high mortality if CCD is misdiagnosed or delayed, as in this case, 2 siblings passed away prior final diagnosis.

### Conflicts of interest

The authors declare that they have no conflict of interest regarding the publication of this case report.

### Funding

None.

### Consent for publication

Written consent was obtained from the parents of the patient.

### Ethical approval

Ethical approval is not required at our institution to publish an anonymous case report.

## Author details

Mohammed AlMesaibli[1], Tuqa A. AlSinan[2], Adeeba Sajid[3], Madiha Jamal[3], Tehreemah Raziq[3]

1. Section of Pediatric Surgery, Department of Surgery, King Saud University Medical City, Riyadh, Saudi Arabia
2. Department of Pediatric Surgery, Prince Sultan Military Medical City, Riyadh, Saudi Arabia
3. College of Medicine, Alfaisal University, Riyadh, Saudi Arabia


## References


1. Di Meglio L, Castaldo G, Mosca C, Paonessa A, Gelzo M, Esposito MV, et al. Congenital chloride diarrhea clinical features and management: a systematic review. Pediatr Res. 2021 Jul;90(1):23–9. https://doi.org/10.1038/s41390-020-01251-2
2. Kamal NM, Khan HY, El-Shabrawi MH, Sherief LM. Congenital chloride losing diarrhea: a single center experience in a highly consanguineous population. Medicine (Baltimore). 2019 May;98(22):e15928. https://doi.org/10.1097/MD.0000000000015928
3. Konishi K, Tatsuki Mizuochi, Yanagi T, Watanabe Y, Kazuhiro Ohkubo, Shouichi Ohga, et al. Clinical features, molecular genetics, and long-term outcome in congenital chloride diarrhea: a Nationwide Study in Japan. J Pediatr. 2019 Nov 1;214:151–7. https://doi.org/10.1016/j.jpeds.2019.07.039
4. Elrefae F, Elhassanien AF, Alghiaty HA. Congenital chloride diarrhea: a review of twelve Arabian children. Clin Exp Gastroenterol. 2013;(Jun)6:71–5. https://doi.org/10.2147/CEG.S40620
5. Sadagah LF, Makeen AZ, Kotbi ET. Diagnostic challenge of congenital chloride diarrhea and ulcerative colitis overlap in an adult misdiagnosed with bartter syndrome: case report and literature review. Am J Case Rep. 2022 Jul;23:e936715. https://doi.org/10.12659/AJCR.936715
6. Wani AM, Janhan N, Hussain WM, Fatani MI, Hemdi M, Imam A, et al. Congenital cataract and congenital chloride diarrhoea--a unique combination and antenatal diagnosis. Case Reports. 2009 Aug 17;2009(aug17 1):bcr0420091775–5. https://doi.org/10.1136/bcr.04.2009.1775
7. Lindberg E, Moller C, Kere J, Wedenoja S, Anderzén-Carlsson A. Congenital chloride diarrhea and pendred syndrome: case report of siblings with two rare recessive disorders of SLC26 family genes. BMC Med Genet. 2020 Apr;21(1):79. https://doi.org/10.1186/s12881-020-01023-z
8. Çelik AT, Barış Z, Aydemir Y, Kocagil S. Late diagnosis of congenital chloride diarrhea mimicking hirschsprung's disease [Internet]. Clin Pediatr (Phila). 2024 Feb;9922824 1228116:99228241228116. [cited 2024 Mar 31] Available from: https://pubmed.ncbi.nlm.nih.gov/38303675/ https://doi.org/10.1177/00099228241228116
9. Cheng Q, Huang C. Prenatal diagnosis of congenital chloride diarrhea: a case report [Internet]. Arch Argent Pediatr. 2024 Jun 1;122(3):e202310167. [cited 2024 Mar 31] Available from: https://pubmed.ncbi.nlm.nih.gov/38019900/


## Summary of the case

| | | |
|---|---|---|
| 1 | **Patient (gender, age)** | Newborn baby boy |
| 2 | **Final diagnosis** | Congenital Chloride Diarrhea (CCD) |
| 3 | **Symptoms** | Passing urine through the anal canal |
| 4 | **Medications** | Fluid resuscitation |
| 5 | **Clinical procedure** | None |
| 6 | **Specialty** | Pediatrics, Pediatric Surgery |