Journal of
Medical Case Reports

CASE REPORT

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Congenital scrotal agenesis with bilateral cryptorchidism: a case report

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Abstract

Background The disorder known as congenital scrotal agenesis is an extremely rare condition. In the literature, 11 cases of congenital scrotal agenesis or absence have been reported thus far. Most of them are bilateral and contain cryptorchidism. In a rare case of bilateral cryptorchidism, we report a 36-day-old Afghan infant who presented to Maiwand Teaching Hospital with fever and vomiting due to infection and congenital scrotal agenesis.

The diagnosis of these anomalies was made using clinical and laboratory results, with the clinical characteristics being recorded during the sixth week of life. After receiving therapy for sepsis, he was referred to the pediatric surgical department in good health. Given that the parents of the congenital scrotal agenesis reference case were consanguineous, there may have been a genetic component to the development of the condition.

Case presentation We describe a rare instance of bilateral cryptorchidism, with congenital scrotal agenesis in a 36-day-old Afghan baby that had come to Maiwand teaching hospital for fever and vomiting on the basis of sepsis.

The clinical characteristics were noted during the sixth week of life and clinical and laboratory results were used to determine the diagnosis of these anomalies. He was referred to the pediatric surgery department in good health following the treatment for sepsis. Given that the reference case of congenital scrotal agenesis had consanguineous parents, a link to genetics may have contributed to the disease's emergence.

Conclusion Congenital scrotal agenesis is an incredibly uncommon condition of urological congenital disorder. To date, the literature has reported 11 cases of congenital scrotal agenesis or absence, most of which are bilateral and involve cryptorchidism. This case presents another human being that suffers congenitally from scrotal agenesis with bilateral cryptorchidism who was born in Kabul city of Afghanistan from a multigravida mother who has positive consanguinity to her husband.

Keywords Bilateral undescended testis, Congenital scrotal agenesis, Cryptorchidism

Background

Congenital scrotal agenesis (CSA) is a rare urogenital tract deformity in which the bilateral testes are present in a cryptorchid or ectopic position and the scrotal rugae are completely absent from the perineum between the penis and the anus. With cryptorchidism, scrotal agenesis is the absence of scrotal skin. Both abnormalities may appear alone or in combination with other defects.

The disorder known as CSA is extremely rare. In the literature, 11 cases of congenital scrotal agenesis or absence have been reported, and thus far the majority are

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bilateral and include cryptorchidism. Only two cases of unilateral scrotal agenesis without cryptorchidism have been documented. This is another instance of bilateral scrotal agenesis combined with bilateral cryptorchidism. While CSA is incredibly uncommon, cryptorchidism in children is a prevalent condition. CSA has thus far been documented in 11 cases according to the literature [1–5]; 2 of these cases are unilateral, the rest are bilateral, and cryptorchidism is present in the majority of them. Our institution recently provided care for a child who had bilateral congenital scrotal agenesis and bilateral cryptorchidism. He was sent to the pediatric surgery for a particular correction of the issue after receiving treatment for his illness.

Case presentation

A 36-day-old Afghan male baby weighing 2.7 kg who was born at 38 weeks of gestation to a 35-year-old multigravida mother (having had 8 live births and 2 stillbirths) by vaginal delivery at a local hospital in 2023 was presented. The mother was in good health and had not been exposed to any teratogen medications or radiation during her pregnancy. She had never used any contraceptive medication. The parents were consanguineous. Her husband was the nephew of her aunt. The child's siblings do not have any congenital anomalies.

He was referred to our department at day 36 of life because of poor sucking, fever, vomiting, and irritability, with a suspected diagnosis of sepsis. During a physical examination, the patient had sluggish primitive reflexes, rectal temperature of 39.9 °C, respiratory rate of 69 breaths per minute, heart rate of 148 beats per minute, and minor chest indrawing with a 90% oxygen saturation level. Hemoglobin levels of 17.4 g/dl, total leukocyte count of 16,200/mm^3 (polymorphs 78.1%, lymphocytes 17.4%%, eosinophil 2%, and monocytes 2%), platelet count of 386,000/mm^3, and C-reactive protein levels of 24 mg/dl were all found during blood tests. On genitourinary system assessment, testes were absent on both sides and the scrotum did not exist. (Fig. 1).

The diagnosis of sepsis was made using abnormal clinical symptoms, leukocytosis, and an increased level of C-reactive protein. An abdominal ultrasound revealed congenital scrotal sac absence and bilateral cryptorchidism. During the first and second days, the patient received nasogastric (NG) tube feeding, and intravenous antibiotics consisting of cefotaxime and ampicillin.

Doppler echocardiography was performed to rule out any possible associated heart problems; however, it found no abnormalities. The patient was ultimately identified as having sepsis, bilateral cryptorchidism, and bilateral congenital scrotal agenesis. Since his hospitalization, he has been stabilized and is breastfeeding well without requiring NG tube feeding or breathing support. In addition, he was referred to the pediatric surgery department to receive particular support for his congenital defect.

Discussion and conclusion

Congenital scrotal agenesis (CSA) is an incredibly uncommon condition of urological congenital disorder. In total, 11 examples of congenital scrotal agenesis or absence have been documented in the literature thus far; the majority being bilateral and including cryptorchidism [2]. This case is another human being that suffers congenitally from scrotal agenesis with bilateral cryptorchidism who was born in Kabul city of Afghanistan from a multigravida mother who has positive consanguinity to her husband.

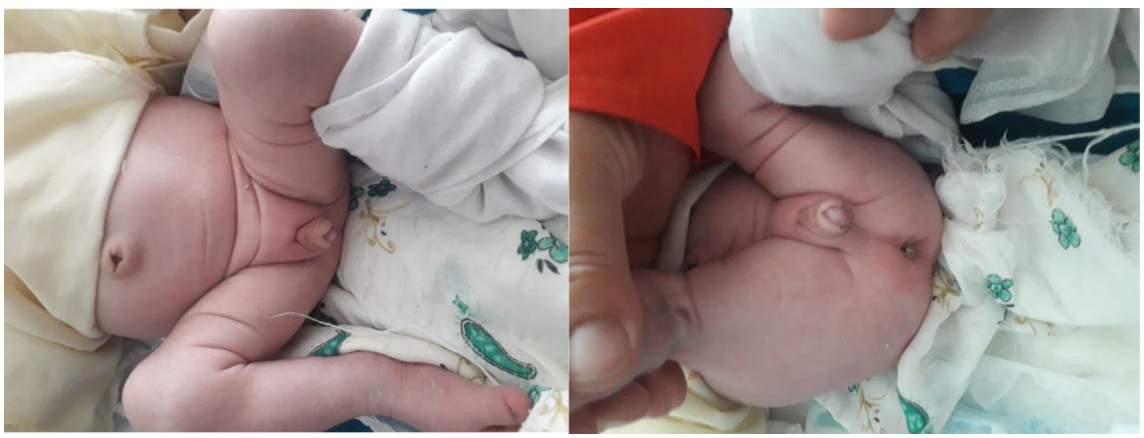

Fig. 1 Picture showing absence of scrotum and testis

This condition arises due to aberrations during the differentiation of the genital tubercle and labioscrotal folds in early embryogenesis. Normally, the scrotum develops from the labioscrotal folds under the influence of dihydrotestosterone. Disruption of this process, whether due to genetic mutations, hormonal imbalances, or environmental exposures, may result in scrotal agenesis [3].

Management of congenital scrotal agenesis is multifaceted and often involves a combination of surgical reconstruction and psychosocial support. The timing of surgical intervention remains a matter of debate; however, early reconstruction may minimize psychological distress and improve quality of life, but long-term outcomes remain poorly documented [4].

This case highlights the importance of a multidisciplinary approach, involving pediatricians, urologists, endocrinologists, and psychologists. Regular follow-up is crucial to monitor for complications, including potential infertility or hormonal imbalances.

A comprehensive review of the literature revealed only a limited number of cases, indicating a significant gap in understanding the etiology, optimal management, and long-term outcomes of this condition. Further research is warranted to elucidate the genetic and environmental factors contributing to congenital scrotal agenesis and to develop standardized treatment guidelines [1–10].

Acknowledgements
Not applicable.

Author contributions
Mohammad Sharif Sediqi, Abdulwali Wali, and Khatera Wali contributed to conception and design as well as the acquisition, analysis, and interpretation of data. Khesrow Ekram and Abdulhai Wali contributed to acquisition and interpretation of imaging data. Mohammad Sharif Sediqi documented the patient's status and contributed to analysis and interpretation of data. All authors read and approved the final manuscript.

Funding
No funding was received.

Availability of data and materials
Not applicable.

Declarations

Ethics approval and consent to participate
We complied with ethical requirements and obtained informed consent from the patient.

Consent for publication
Written informed consent was obtained from the patient's legal guardian for publication of this case report and any accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal.

Competing interests
The authors declare that they have no competing interests.

Received: 18 July 2023 Accepted: 1 April 2025
Published online: 06 May 2025

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