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Clinical Image of Aphallia: A Rare Congenital Anomaly

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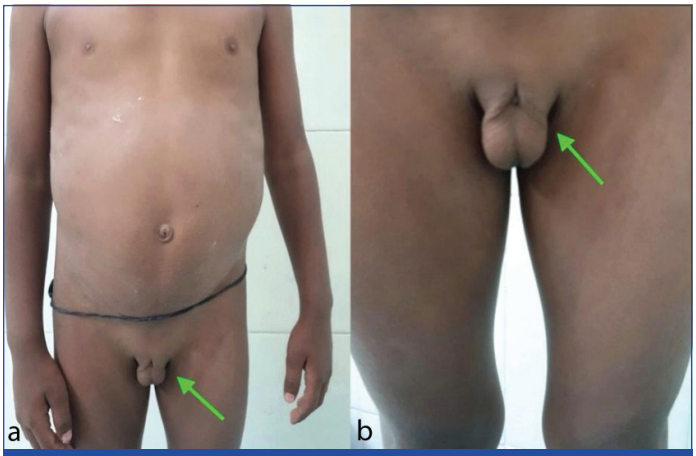
A six-year-old boy with a karyotype of 46XY presented to the outpatient department with congenital absence of the penis, known as aphallia [Table/Fig-1]. He was born from an uncomplicated pregnancy, with no family history of congenital anomalies or consanguinity. Upon further history-taking and physical examination, it was discovered that he was passing urine from an external urethral opening located in the perineum. A local examination revealed a total absence of the penis, and bilateral testes could not be palpated in the scrotum. Apart from this, there were no other identifiable external abnormalities. An abdominal and inguinoscrotal ultrasound showed left kidney pyelectasis, and the right testis was found to be undescended, located in the inguinal canal, while the left testis was present at the neck of the scrotum. Doppler ultrasound indicated internal vascularity in both testes. Due to these findings, the parents were counselled regarding the nature of the condition, and the patient was ultimately referred to a higher centre for further management by a specialist in Disorders of Sexual Differentiation (DSD) regarding the case of aphallia.



[Table/Fig-1]: Clinical images (a and b) of the patient with aphallia.

Aphallia, or complete penile agenesis, is defined as the congenital absence of the penis. It is an exceedingly rare condition, with a reported prevalence of just one in 10 to 30 million live births and fewer than 100 documented cases in the medical literature to date [1]. The primary pathological cause of aphallia is the absence or impairment of genital tubercle development during the first four weeks of embryonic development. In instances where aphallia is associated with more severe malformations, the underlying issue may arise from defects in cloacal differentiation during early embryogenesis, specifically as a defect in blastogenesis. A diagnosis of aphallia is characterised by the absence of the phallus, a male karyotype, and a normally developed scrotum containing normal testes [2]. It frequently occurs alongside other congenital anomalies, including kidney agenesis or cystic kidney, horseshoe kidney, urinary reflux, prostate agenesis, skeletal and neural disorders, annular pancreas, clubfoot, and cardiac problems, which can lead to life-threatening complications [3].

A notable genitourinary complication was reported in a case study by Malik MA et al., involving a nine-year-old boy who presented with fever, vomiting, drowsiness, aphallia, and uraemic urosepsis. He underwent bilateral percutaneous nephrostomy for severe hydronephrosis and was started on broad-spectrum antibiotics, leading to clinical improvement. However, despite recommendations for further surgical procedures, including appendico-vesicostomy and closure of vesico-rectal communication, the parents refused treatment and left the hospital without follow-up [4].

The treatment of this genetic abnormality requires a multistage and multidisciplinary approach involving paediatricians, paediatric surgeons, geneticists, endocrinologists, and psychologists. The primary management focuses on addressing life-threatening associated malformations, after which elective surgeries, such as gender reassignment or reconstructive surgeries, can be conducted.

A case report by Gupta A and Gupta H detailed the situation of a six-month-old boy diagnosed with this condition, emphasising the difficult decisions his parents faced regarding gender reassignment amid societal pressures. After receiving counselling on treatment options for their child with aphallia, the family considered two main approaches. The first option was female gender reassignment, which involved bilateral orchiectomy and urethral-perineal transposition, with plans for future feminising genital reconstruction. The second option, male phallic construction, was ultimately deemed less favourable due to the high failure rate in achieving erectile function and the potential for sexual difficulties later in life. Given these considerations, the family chose female gender reassignment, as it offered more favourable prospects for normal sexual function compared to the risks associated with male phallic construction [5]. Early surgical intervention is crucial, and parents should receive thorough counselling on all treatment options, including their advantages and disadvantages, to make an informed decision.

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