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## Case Report

# A rare case report of aphallia with hypoplastic kidney and vesicorectal fistula ☆

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## ABSTRACT

Aphallia or penile agenesis is a rare case of the genitourinary system which has an association with upper urinary tract disorder. Failure of fetal genital tubercle formation in the embryonic period is the cause of this disorder. The incidence of aphallia according to previous studies is about 1 in 10–30 million births. We report a case of a child with the absence of the penis with associated kidney hypoplasia and vesicorectal fistula.

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## Introduction

Aphallia is an extremely rare congenital malformation of unknown cause. The incidence is reported in the literature to be 1 in 10–30 million live births [1]. Aphallia is an anomaly during the fourth week of embryonic development, which accounts for its frequent association with related malformations. It is related to the result of the non-formation of the genital tubercle or its failure to develop. Its diagnostic is easy at birth, as a penis is absent [2]. More than half of these patients have associated anomalies including genitourinary (54%) and gastroin-

testinal tract anomalies and developmental defects of the caudal axis [3].

## Case presentation

We introduce a 14-month-old baby who was born in this center without a penis. The pregnancy that led to his birth was at term. The baby was born with uncomplicated vaginal delivery. His birth weight was 2700 g and his parents also were not related. In the history taken from the mother, she

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**Fig. 1 – Absence penis, well-formed scrotum, and descended testes.**

mentioned only the use of propranolol to control heart rate. At the postnatal examination, the baby was in a good general state and penis was absent and urinary meatus was not detected and the patient passed urine from the anus, the scrotum was developed and both testes were palpable (Fig. 1).

Ultrasound and CT scan with IV contrast were requested for further evaluation of the patient. Initial ultrasound of the abdominopelvic showed hypoplasia of LT kidney with mild stasis and no female pelvic organs were seen in the pelvic cav-

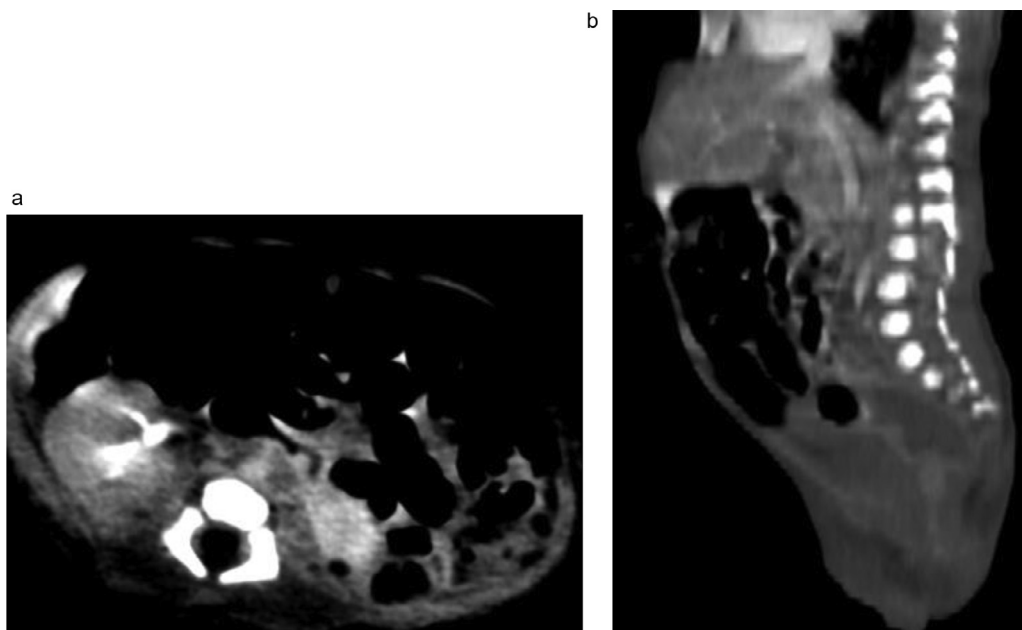
ity. Both testicles and epididymides were normal shape and echogenicity and located in the scrotum. CT scan with IV contrast revealed hypoplasia of LT kidney with reduced secretion from LT kidney, and also small vesicorectal fistula (Fig. 2).

At the last ultrasound performed at the age of 1 year and 2 months, LT kidney was not detected and mild diffuse wall thickening of urinary bladder was seen (Fig. 3). A rectal enema with water-soluble contrast was also performed at the age of 1 year and 2 months and revealed a vesicorectal fistula (Fig. 4).

## Discussion

Penile agenesis (aphallia) is an extremely rare congenital defect characterized by the complete absence of the penis in a child with a male karyotype 46XY [4]. Aphallia is thought to result from the non-formation or failure of development of the fetal genital tubercle or failure in caudal migration of the urogenital sinus between the third and sixth weeks of embryonic development [5].

Evaluation of the patient with aphallia should include genetic assessment and evaluation for associated malformations. Commonly associated malformations include genitourinary anomalies (renal agenesis, hypoplastic kidneys, hydronephrosis, vesicourethral reflux, hypoplastic bladder, vesicocolic fistula, and agenesis of the prostate and seminal vesicles) and gastrointestinal malformations (anteriorly placed anus, tracheoesophageal fistula, and annular pancreas). Other associated malformations include pigeon chest, hemivertebrae, club feet, shortened forearms, accessory earlobes, and low-set ears [6]. In our case, hypoplasia of the LT kidney was detected.



**Fig. 2 – (A) Axial CT scan reveals the small size of LT kidney with decreased secretion rather RT kidney in excretory phase suggestive of LT kidney hypoplasia. (B) Sagittal CT scan shows vesicorectal fistula. (C) Coronal CT scan shows vesicorectal fistula. (D) Excretory phase of CT scan, 3D reconstruction of the collecting system shows nonvisualization of the left kidney.**

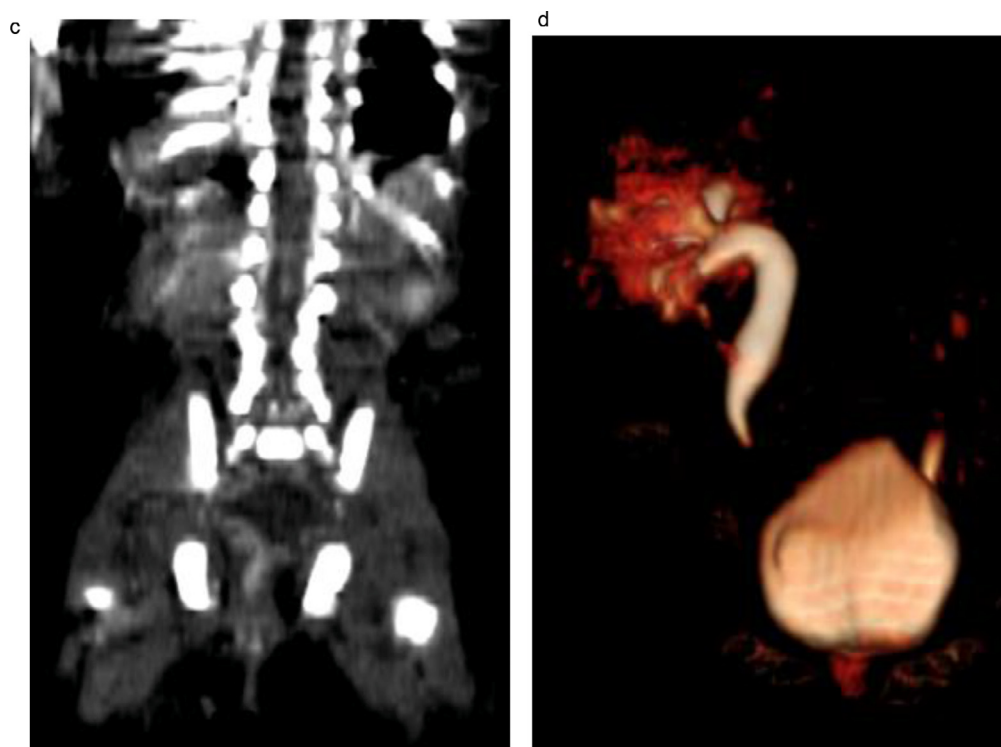
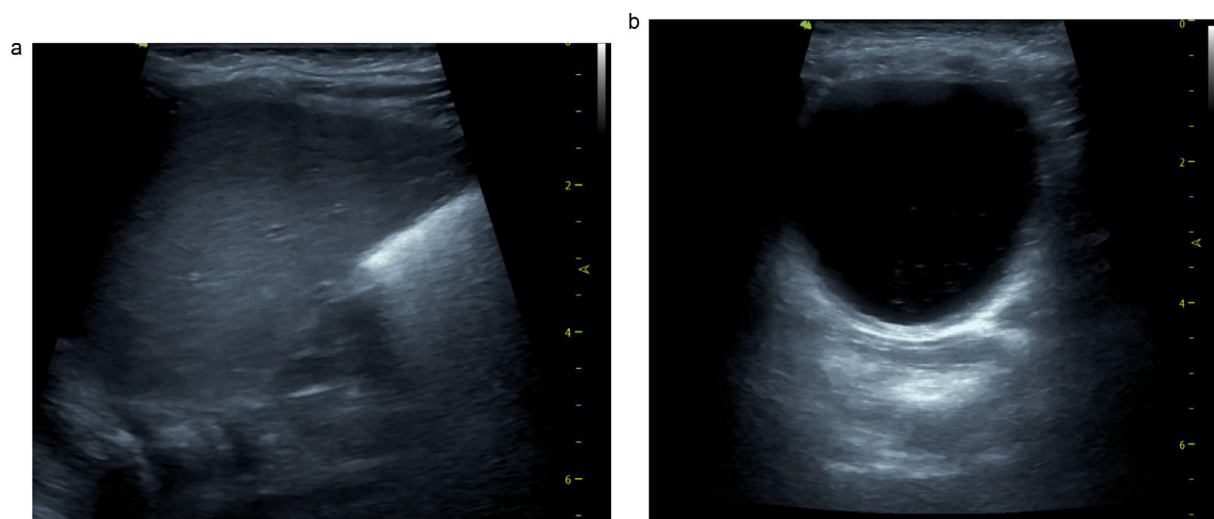


Fig. 2 – Continued



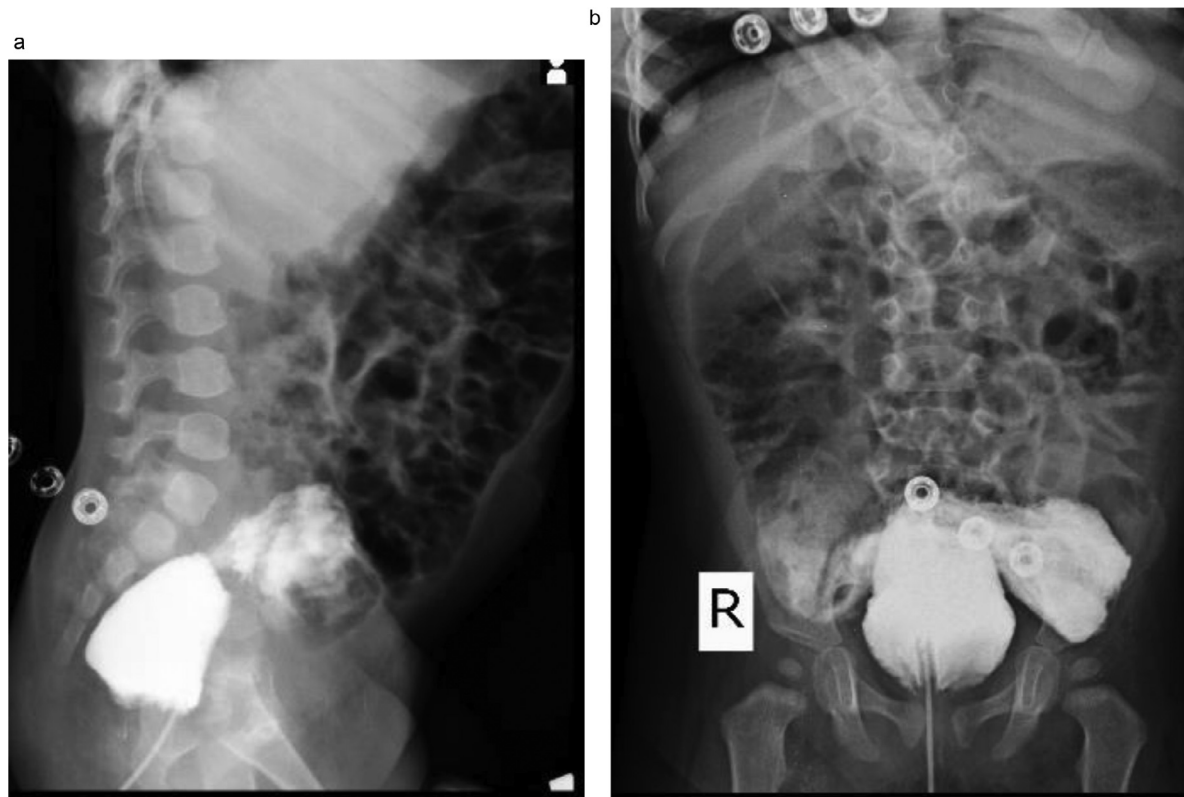
**Fig. 3 – (A) Last abdominal ultrasound shows the absence of LT kidney. (B) Last pelvic ultrasound reveals mild diffuse wall thickening of UB.**

In most cases, aphallia is associated with communication of the urinary tract and rectum, which can be at the anal verge or higher as presented in this case. Skoog and Belman proposed a classification system for penile agenesis where it has been emphasized that the position of the urethral opening has a bearing on the prognosis. A more proximal urethral opening results in a more significant number of associated anomalies and higher mortality [3].

Aphallia has been reported among diabetic mothers. But urogenital system abnormalities are more prevalent among

controlled diabetic mothers [7]. In our case, the baby's mother had no history of gestational diabetes.

In penile agenesis patients, early gender reassignment and feminizing genitoplasty are recommended. In the past, multiple operations were carried out to form feminized external genitalia. Glüer et al. performed a definite genital reconstruction in a neonate using the posterior sagittal approach. The operation included bilateral orchiectomy, urethral reconstruction, sigmoid vaginal replacement, and formation of the labia. Also, estrogen therapy at the age of puberty, along with psy-



**Fig. 4 – (A) Rectal enema lateral view shows vesicorectal fistula. (B) Rectal enema AP view shows vesicorectal fistula.**

chological monitoring is recommended. They emphasize the urethral hypotrophy to be the main challenge in this anomaly and conclude that immediate complete reconstruction is possible and should be performed in patients with this delicate condition [8].

When the diagnosis is late, or the operation was not done in early life for any causes, phallus reconstruction and urethral reconstruction is recommended to minimize psychological trauma for both the child and parents. Chibber et al. reported a patient with aphallia who presented at the age of 16 years and was treated with phallus reconstruction and urethral reconstruction [9].

### Patient consent

We confirm that informed consent was obtained from parents of the patient described in this report.

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