

## Case report

# A Rare Case of Aphallia with Right Kidney Hypoplasia and Left Kidney Dysplasia

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## Abstract

Aphallia or penile agenesis is an extremely rare congenital anomaly with an estimated incidence rate of 1 in 10 to 30 million births. We report a rare case of aphallia with right kidney hypoplasia and left kidney dysplasia in a 10-day old Iranian-Azeri male. The patient had creatinine rise and renal failure due to dysplastic left kidney and hypoplastic right kidney and expired on fifth day of admission. There were only six cases of renal malformation associated with aphallia in the literature review. Three of the cases were complicated by Potter sequence and one of them was accompanied by chronic renal failure. Our case had a unique presentation because of bilateral renal malformation and subsequent renal failure without the Potter sequence.

**Keywords:** Aphallia, gender, kidney dysplasia, kidney hypoplasia, penile agenesis

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## Introduction

Aphallia or penile agenesis is a rare congenital defect in which the phallus is absent. It is an extremely rare disorder with potential urogenital and psychological consequences.<sup>1</sup> It has an estimated incidence rate of 1 in 10 to 30 million births.<sup>2,3,4</sup> Most of the patients have 46 XY karyotypes<sup>5</sup> and therefore, early gender reassignment is recommended.<sup>3,5</sup> More than half of aphallia cases have associated anomalies including caudal axis, cardiovascular, genitourinary and gastrointestinal anomalies.<sup>5,6</sup> Here, the authors report a case of aphallia associated with right kidney hypoplasia and left kidney dysplasia.

## Case Reports

A neonate (3450 gr weight; 50 cm height) was born by normal vaginal delivery (NVD) to a 37-year-old woman (Gravid 2, Para 2) in the 38<sup>th</sup> week of pregnancy in Maragheh, Eastern Azerbaijan, Iran. The mother did not have any prenatal sonographic evaluation but there were no prenatal or peripartum difficulties. There was no history of teratogen ingestion. There was no family history of congenital anomalies, especially urogenital anomalies. The neonate had aphallia and was referred to Children's hospital, Tabriz,

Iran for urgent care.

The patient was 12 hours old on admission and had meconium passage mixed with urine. The physical examination demonstrated a well developed male, except for complete absence of a phallus or corporal tissue. The scrotum was normal and the neonate had bilaterally descended testes (Figure 1). Anus was placed normally and the urethral opening was not visible anywhere in the perineum. The neonate was in good condition and did not have any clinical evidence of other associated anomalies; however, he had creatinine rise on routine laboratory examinations and daily pediatric nephrology visit was ordered to monitor the kidney function. The urine passage was through the rectum by a urethrorectal fistula located at the exit site of the rectum. Ultrasound examination revealed a small sized right kidney (18 × 20 mm) without any stones or hydronephrosis. Left kidney was also dysplastic and a cystic mass (17 × 29 mm) was detected within the dysplastic kidney (Figure 2). The ureters and bladder were normal. The patient had further creatinine rise and renal failure due to his dysplastic left kidney and hypoplastic right kidney on the following days. The parents of the neonate did not give consent for the treatment of the condition. Finally, the neonate continued to have creatinine rise and glomerular filtration rate (GFR) decline and expired with severe renal failure on the fifth day of admission.

## Discussion

Aphallia is an extremely rare congenital anomaly with profound urogenital and psychosocial consequences.<sup>1</sup> Approximately 60 cases have been reported as of 1989<sup>1</sup> and 75 cases as of 1997.<sup>7</sup> Aphallia is with or without associated anomalies with a range of caudal axis, cardiovascular, genitourinary and gastrointestinal anomalies.<sup>5</sup> It has been estimated that more than half of aphallic cases have associated anomalies<sup>5,6</sup> and the type and severity of these anomalies surely affect the survival of the patients. Proximal meatus increases both the incidence of other anomalies and the

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**Figure 1.** Photograph illustrating aphallia with a normal scrotum.



**Figure 2.** Ultrasound findings of the presented case.

number of neonatal deaths.<sup>1</sup>

We present a rare case of aphallia associated with right kidney hypoplasia and left kidney dysplasia. This case is of potential use and clinical importance because of its unique presentation with bilateral renal malformation and subsequent renal failure. Attie<sup>8</sup> reported a case of aphallia with congenital concealed penile agenesis and congenital absence of the left kidney and ureter in 1961. O'Connor *et al.*<sup>9</sup> reported another case of unusual aphallia associated with agenesis of the kidneys and urethra leading to fatal oligohydramnios sequence with pulmonary hypoplasia in 1993. Another case of aphallia with renal involvement was reported by Arai *et al.*<sup>10</sup> in 2001 which was complicated by Potter sequence. They reported a case of penile agenesis complicated by multicystic dysplastic kidneys and urethral agenesis, which resulted in oligohydramnios, pulmonary hypoplasia and neonatal death. Arai *et al.*<sup>10</sup> suggested that cases of penile agenesis complicated by Potter sequence with urethral agenesis should be differentiated from those with ectopic urethral opening. In addition, Dursun *et al.*<sup>11</sup> reported another case of aphallia complicated by bilateral multicystic renal dysplasia with Potter sequence. In another case reported by Ciftci *et al.*,<sup>12</sup> a child with aphallia expired due to chronic renal failure. Gérard-Blanluet *et al.*<sup>13</sup> presented a case of aphallia with other associated anomalies including imperforate anus, bilateral renal dysplasia and complete right lung agenesis and rib segmentation anomaly. Our case is differentiated from the cases reported by O'Connor *et al.*,<sup>9</sup> Arai *et al.*,<sup>10</sup> and Dursun *et al.*<sup>11</sup> in that the present case did not undergo the Potter sequence. Potter sequence is mostly believed to result from a renal or urologic abnormality such as bilateral renal agenesis, cystic dysplasia, obstructive uropathy etc.<sup>14</sup> In the three cases reported by O'Connor *et al.*,<sup>9</sup> Arai *et al.*,<sup>10</sup> and Dursun *et al.*,<sup>11</sup> penile agenesis was associated by bilateral renal malformation and renal failure with subsequent Potter sequence. These cases were accompanied by fatal oligohydramnios sequence with pulmonary hypoplasia. These cases suggest a distinct relationship between aphallia and renal malformation, and thus, Potter sequence can complicate the affected neonates due to renal failure. The absence of Potter sequence in our case is a

unique presentation that attracts intense attention and clinical consideration. The present case was associated with a dysplastic left kidney with a cystic lesion and the right kidney was also hypoplastic. The probable reason for the absence of Potter sequence in this case could be the incomplete failure and gradual function loss of the right kidney prenatally and during the neonate's five-day life. The pattern of creatinine rise, GFR decline and renal failure was also compatible with this explanation. The neonate also had urine passage mixed with meconium and this could also be due to the remaining function of the hypoplastic right kidney.

Aphallia is a complex urogenital and psychosocial problem and the treatment needs a complete assessment of clinical and psychosocial factors.<sup>2</sup> It is suggested that opposite gender should not be assigned in patients affected by penile agenesis, who are better raised according to their karyotype and hormonal production.<sup>15</sup> Besides, a number of authors believe that treatment demands feminizing genitoplasty in most patients.<sup>16</sup> Definitive phalloplasty in adults may yield good results.<sup>15</sup> There are different surgical techniques used for patients suffering from aphallic disorders as De castro to create neophallus<sup>17</sup> or reconstruction of urethra from pedicled skin flap mixed with buccal mucosa graft.<sup>18</sup> Our case expired on his fifth day of life and thus we did not conduct any procedures for treatment and management of his aphallia.

In conclusion, Aphallia is an extremely rare congenital anomaly with or without associated anomalies. We reported a rare case with right kidney hypoplasia and left kidney dysplasia. There were four cases of renal anomalies associated with aphallia in the literature review. Three of the cases were complicated by Potter sequence. Our case had a unique presentation because of bilateral renal malformation and subsequent renal failure without the Potter sequence. This is surely of potential clinical consideration.

### Consent section

Written informed consent was obtained from the patient's legal guardian for publication of this manuscript and accompanying images.

## Competing interests

The authors declare that they have no competing interests.

## Authors' contribution

SA, HA and RR contributed to the preparation of the manuscript. SB, DB and MABF contributed to the literature review and discussion. SZ contributed to all parts of manuscript preparation, case presentation and intellectual interpretation.

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