www.jpnim.com Open Access eISSN: 2281-0692
Journal of Pediatric and Neonatal Individualized Medicine 2023;12(1):e120126
doi: 10.7363/120126
Received: 2022 Dec 13; revised: 2023 Jan 23; accepted: 2023 Feb 06; published online: 2023 Apr 19

Case report

Diphallia as the first sign of a polymalformative syndrome

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Abstract

Diphallia or penile duplication is a rare anomaly, with an estimated prevalence of 1 in every 5.5 million live births. It may present alone or associated with other penile or abdominal malformations, such as renal and anorectal anomalies. At the genetic level, defects in the homeobox genes may be involved.

We report a case of a 4-year-old boy, born at term, with adequate for gestational age anthropometrics, after a medically supervised pregnancy with normal prenatal ultrasounds. He exhibited two completely developed and structurally normal penile glands with urinary emission from the ventral penis, a right gluteus hypertrophy with a left side deviation of the internal groove with a well-positioned anus.

Follow-up exams confirmed the presence of two complete penises (complete diphalia), a normal sized single bladder connected with two urethras (only one of which was working), and no valves or vesicoureteral reflux. The 99mTc-DMSA scintigraphy showed two kidneys, with a left uptake of 94.41%. The karyotype was normal and no cytogenetic test was conducted yet at the parents' request.

During these 4 years no urinary infection was reported and normal kidney function was confirmed in the last analytical panel.

He also exhibits a global developmental delay, mostly in the language area, and is currently not enrolled in kindergarten.

The surgical approach consisted on the excision of the accessory penis (with no functional urethra) and was conducted without complications.

We describe a rare case of a complex polymalformative syndrome, where early and appropriate diagnosis and treatment are a major challenge and depend on the commitment of a multidisciplinary team. One has to ensure normal urinary and reproductive functions, while balancing the psychological impact, as the child gets older and more aware of physical differences.

Keywords

Diphallia, congenital malformation, urogenital abnormalities, CAKUT, polymalformative syndrome.

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How to cite

Correia C, Correia MR, Vaz R, Ribeiro-Castro J, Peixoto S. Diphallia as the first sign of a polymalformative syndrome. J Pediatr Neonat Individual Med. 2023;12(1):e120126. doi: 10.7363/120126.

Case presentation

We report a case of a 4-year-old boy, born at term by vaginal delivery, weighting 3,480 g and measuring 51 cm (adequate for gestational age). He is the third son of a 30-year-old-mother with no relevant family history. This was a medically supervised pregnancy complicated with gestational diabetes (requiring oral antidiabetic drugs) and tobacco consumption (2 per day), with apparently normal prenatal ultrasounds.

At birth, his Apgar scores at 1 and 5 minutes were 9 and 10, respectively. He exhibited two completely developed and structurally normal penile glands with urinary emission from the ventral penis (**Fig. 1**). In addition, a right gluteus hypertrophy with a left side deviation of the internal groove and a well-positioned anus was noted (**Fig. 2**). No other macroscopic malformations were identified at the time. Clear urine was passed in the first 24 hours of life.

A penile ultrasound (US) confirmed the presence of two complete penises with apparent bulb convergence and two structurally normal testicles. An abdominal US showed only one normal-sized left kidney (measuring 5 cm lengthwise) and one bladder, with no other abnormalities.

In the first 24 hours he required supplementation with glucose due to hypoglycemia. There were no other perinatal complications, and the home discharge was uneventful.

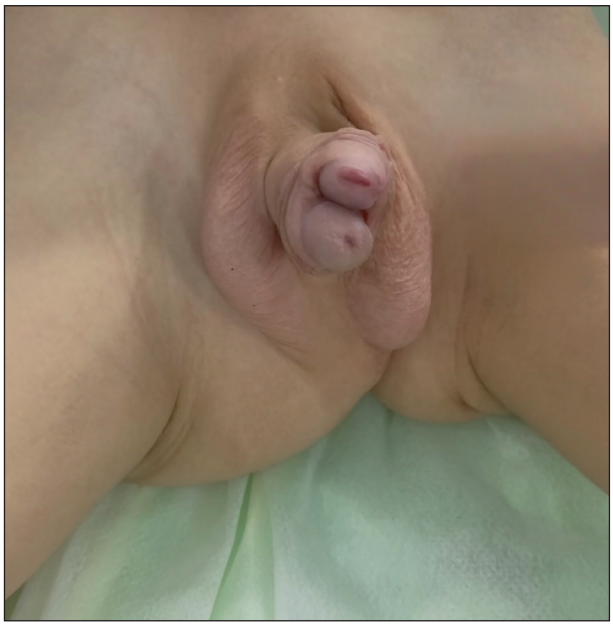

Figure 1. Penile duplication with two phalli disposed in the sagittal plane, both with apparent urethral openings.

Figure 2. Penile duplication and left side deviation of the internal groove.

Posteriorly, a cystourethrography revealed a normal-sized single bladder connected to two urethras (only the ventral one was working) and no valves or vesicoureteral reflux. 99mTc-DMSA scintigraphy showed two kidneys, with a left uptake of 94.41% versus 5.59% on the right.

The patient's karyotype was 46XY, and no cytogenetic tests were conducted at the parents' request.

A limitation in the abduction of the right eye was noted in the first year of life and evaluated in an ophthalmology consultation, with suspicion of type I Duane syndrome. Ear positioning asymmetry is becoming more apparent with aging.

During these 4 years, no urinary infection was reported, and antibiotic prophylaxis was discontinued at 12 months. Normal kidney function was confirmed in the last analytical panel.

The child is between the 3rd and 15th percentiles of weight, the 3rd percentile in height, and above and parallel to the 97th percentile in head circumference since birth.

Currently (4 years), he exhibits global developmental delay, mainly in the language area, and is not enrolled in kindergarten. He has no sphincter control (neither for urine or feces).

A cerebral magnetic resonance imaging (MRI) scan was conducted, exhibiting only a benign subarachnoid space enlargement and no structural anomalies (particularly malformities or lesions of metabolic and expansive nature).

A pelvic MRI confirmed the perineal dysmorphia, with increased subcutaneous fat on the right side and a left side deviation of the median raphe. It showed a duplication of the cavernous bodies, with two independent penises in juxtaposition on the sagittal plane and individual glans (**Fig. 3** and **Fig. 4**). Only the left penis appeared to have a urethra within the spongy body and no bladder or anorectal malformations were identifiable.

Intra-operative findings revealed no spongy body in the "accessory" penis (right phallus), and cystoscopy confirmed that there was only a blind end urethric dimple in its glans. After degloving, it was also established that the urethra and spongy body on the left phallus were morphologically normal, without fistula to the contralateral one. As such, the right phallus was then carefully dissected and amputated. Patient was discharged on the fourth post-operative day and there were no post-operative complications.

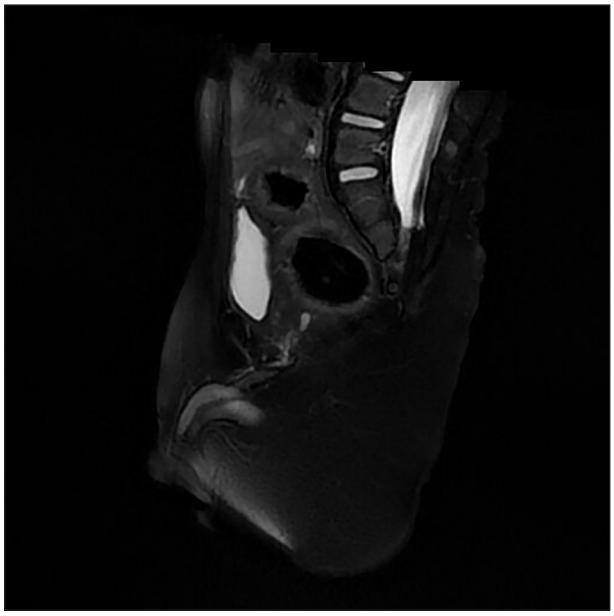

Figure 3. Sagittal section image from the pelvic magnetic resonance imaging (MRI) exhibiting the duplication of the cavernous bodies, with two independent penises in juxtaposition.

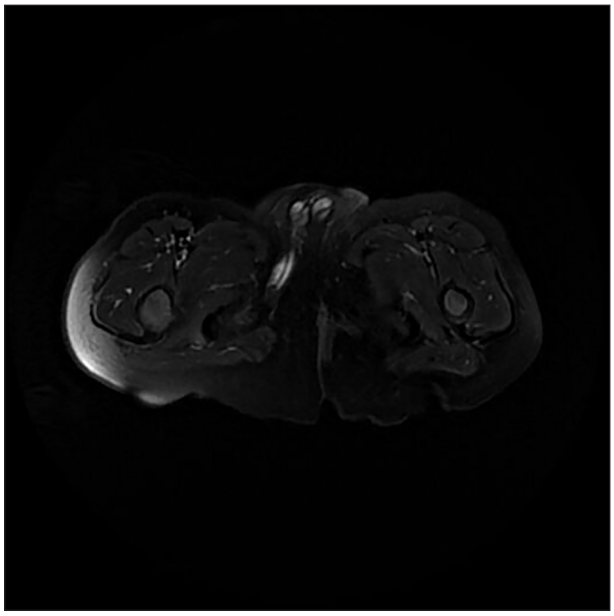

Figure 4. Transversal section image from the pelvic magnetic resonance imaging (MRI) exhibiting the duplication of the cavernous bodies, with two independent penises in juxtaposition.

Discussion

Diphallia or penile duplication is a rare anomaly, with an estimated prevalence of 1 in every 5.5 million live births. The first case was reported in 1609, and since then, just over 100 cases have been reported. It may present alone or associated with other penile or abdominal malformations [1-

4], such as renal and anorectal anomalies, as seen in this case. Other possible associated urogenital anomalies include epispadias in either or both the phalli, exstrophy, or bladder duplication.

According to Schneider [5], diphallia may have three presentations (excluding pseudo-diphallia): duplication of the glans only, bifid diphallia, and complete diphallia, with each penis having two corpora cavernosa and a corpus spongiosum. This is a case of complete diphallia.

In 2017, Jesus et al. [6] focused more in glans duplication, and after conducting a review, suggested a new classification. This group of patients normally presents with esthetic/sexual complaints or obstructed voiding patterns caused by urethral associated abnormalities after pubertal genital development.

Recently, Kendrick and Kimble [7] proposed a new classification system based on the anatomical variants previously described in the literature (review of 87 relevant patients), intended to provide a more detailed description and separate diphallia into specific categories based on the structure of the phallus, the urethral anatomy and the bladder formations. This new system places the focus on the structure of the phallus that is to be kept and the pathway of the urethra present within the most normal phallus.

Embryologically, it is linked to a defect connecting the genital tubercle [8, 9] and urethral folds and seems to occur between the 23rd and 25th days of gestation, probably due to epigenetic mechanisms linked to strong environmental factors. The explanation seems to be either "separation" of the pubic tubercles, in which each phallus has one corporal body and urethra, or "cleavage" of the pubic tubercle, in which each phallus has two corporal cavernous bodies and urethras.

At the genetic level, Karna and Kapur [10] associated diphallia with a chromosomal imbalance caused by t(1.14) translocation (p36.3; q24.3), suggesting that defects in homeobox genes, considered the driver genes of the differentiation process, may be involved.

We describe a rare case of a complex polymalformative syndrome, where early and appropriate diagnosis and treatment are a major challenge and depend on the commitment of a multidisciplinary team. The unique and significant aspects of the present case are several. First, it can be characterized as a complete penile duplication, which accounts for only one-third of all reported and extremely rare diphallia cases. Second, aside from the renal hypo-dysplasia, no other anomaly often associated with diphallia, such as duplication of the bladder, urethra and colon, bladder or cloacal exstrophy, anorectal malformations, and vertebral deformities, were observed. Third, despite the complex anatomy of the genitourinary system, no urinary infection was reported over almost 4 years.

Summary learning points

- Congenital malformations should always encourage the clinician to investigate further.
- Urogenital abnormalities, despite being common, are a surgical challenge. One has to ensure normal urinary and reproductive functions, while balancing the psychological impact as the child gets older and more aware of physical differences.
- Diphallia cases, specifically, are very rare, ranging from small accessory phallus to complete duplications, commonly with size asymmetry and sagittal juxtaposition, as detailed in our case.
- Complex anomaly associations may only become apparent with age; therefore, extended follow-up is warranted.
- Intervention is typically approached on an individual basis, as no two patients are alike, and surgical treatment is usually undertaken in a stepwise manor, based on urethral patency and functionality.

Consent for publication

Parental approval was granted and signed; nothing in the manuscript refers to the identity of the child.

Declaration of interest

The Authors declare that there is no conflict of interest.

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