

Neonatal Duplicated Penis with Associated Complex Cardiac, Gastrointestinal, Renal, and Musculoskeletal Abnormalities

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Abstract:

Diphallia is an exceptionally rare genitourinary anomaly often associated with caudal duplication syndrome, which poses challenges in understanding its diverse nature. This case report presents a unique manifestation in a male infant born at 36 weeks to a mother with uncontrolled diabetes. The infant exhibited complete duplication of the penis, scrotum, testicles, urethra, bladder, sacrum, and right kidney, alongside major anomalies including Tetralogy of Fallot and lumbosacral spinal dysraphism with lipomyelomeningocele. Minor anomalies included abnormal ear creases, hypospadias, hydronephrosis, rectal-urethral fistulas, and a left club foot. This case represents the first documentation of such a complex constellation of supraumbilical presentation that includes abnormal ear creases and cardiac associations of Tetralogy of Fallot, right-sided aortic arch, and mirror aortic arch vessels.

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Introduction

Diphallia, or duplicated penis, is an extremely rare genitourinary congenital anomaly that can present in newborn infants. Each case is unique in the degree of duplicated penile tissue and other associated congenital anomalies (1–3). Diphallia is usually associated with other genitourinary, gastrointestinal, and other congenital defects such that no two cases are exactly alike (1, 2). The exact etiology of diphallia is still unknown and is multifactorial in nature (1, 3, 4). Diphallia is often associated with caudal duplication syndrome (CDS), a constellation of a duplicated distal GI tract, urinary tract, genital organs, and/or lumbosacral spine, and other associated anomalies (5, 6). Given diphallia's rare and varied nature, additional case reports are necessary to identify risk factors, associated anomalies, appropriate screenings, and required surgical interventions.

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In our case report, we present a unique presentation of diphallia and CDS from an infant boy born at 36 weeks. To our knowledge, this is the first case report with such a unique, complex constellation of multiple congenital anomalies for both CDS and isolated diphallia. This presentation includes previously unreported cardiac associations of Tetralogy of Fallot, right-sided aortic arch, and mirror image branching of aortic vessels.

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Figs. 1A and 1B: Ear anomalies, bilaterally folded helix with abnormal creasing.



1A

1B

Fig. 3A: Left sided defect representing lipomyelomeningocele

Fig. 3B: Left shortened lower extremity and clubbed foot



3A

Case

Our patient is a male infant born to a 36-year-old G₂P₂ woman with poorly controlled Type 2 diabetes, hypertension, and preeclampsia. The prenatal evaluation was significant for a 21-week fetal ultrasound that was concerning for multiple congenital anomalies, including spina bifida, Tetralogy of Fallot, and bilateral clubbed feet. The rest of her prenatal imaging, labs, and care were unremarkable.

The baby was delivered emergently at 36 weeks 6 days via C-Section due to severe maternal hypertension. Multiple obvious congenital anomalies were noted at birth. His physical exam was insignificant for abnormal ear findings, a completely duplicated penis, a duplicated scrotum, gluteal and sacral dimples, and left lower leg clubbing, as seen in **Figures 1–3**. The patient completed multiple imaging studies with abnormal congenital findings summarized in **Table 1**. His genetic evaluation was performed after completing a whole exome sequencing study. The study did not detect relevant sequence variants.

Due to the complex nature of the patient's condition and congenital anomalies, a multidisciplinary approach was required. Con-

Fig. 2A: Completely duplicated penis and scrotum.

Fig. 2B: Duplicated scrotum with abnormal midline scrotal tissue. Midline gluteal cleft with anatomically positioned anal sphincter. Left gluteal dimple without evidence of anal sphincter structures or fistula. Skin tag present



2A



2B



3B

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His prenatal diagnosis of Tetralogy of Fallot was confirmed with a

Table 1. The evaluation included examination under anesthesia, ultrasound, MRI, VCUG, retrograde pyelogram studies, upper GI, and barium enema.

Patient Evaluation for Congenital Anomalies	
HEENT	• Bilateral folded ear helix with abnormal ear creases
Genitourinary	• Complete duplicated penis, urethra, and bladder • Glanular hypospadias of the right penis • Duplicated scrotum with abnormal medial scrotal skin • Complete testicular duplication; 4 testicles in total
Renal	• Ectopic left kidney transversely positioned at midline • Crossed fused renal of the left kidney to the lower pole of the right kidney • Duplicated right kidney • Right grade 2 hydronephrosis • Two rectal, urethral fistulas originating at the left penile urethra bladder neck
Cardiac	• Tetralogy of Fallot • Right-sided aortic arch • Mirror image branching of aortic vessels
Gastrointestinal	• Lateral anal-sacral dimple without fistula • Sacral skin tag
Neurologic	• Tethered, duplicated cord at the level of L4 • Lumbosacral spinal dysraphism with a lipomyelomeningocele and lipomyeloschisis • Spinal syrinx extending from T12
Musculoskeletal	• Lower lumbar spine, sacrum, and coccyx dysgenesis • Duplicated, dysplastic sacrum • Hypoplastic left gluteus with abnormal fatty tissue • Shortened left lower extremity • Left club foot

postnatal echocardiogram. The baby remained stable and did not experience desaturations or tet spells during his time in the NICU. Serial echocardiograms were performed to assess his pulmonary stenosis. He required initiation of propranolol due to worsening pulmonary stenosis gradients. The patient was sent home with close outpatient follow-up with pediatric cardiology. At 4.5 months old, he underwent a PDA ligation and a transannular VSD patch closure. He has recovered very well from this surgery to date. Follow-up echocardiograms showed no residual ventricular level shunting, no right ventricular outflow obstruction, expected free pulmonary insufficiency, and normal biventricular systolic function. His Lasix and propranolol were weaned off by 8.5 months of life. He is expected to have a pulmonary valve replacement between 5 and 15 years, depending on his growth and clinical condition.

Our patient encountered challenges in achieving full feeds and required a G-tube placement before discharge. The presence of vesicoureteral reflux on the left side and bilateral hydronephrosis predisposed our patient to urinary tract infections caused by *Enterobacter* and *Klebsiella oxytoca*, subsequently leading to bacteremia and meningitis. This issue required a prolonged antibiotic treatment, and the infections responded appropriately.

Pediatric Urology advised correcting the recto-urethral fistula around 6 to 12 months, advising prophylactic amoxicillin until repair. They recommended a repeat outpatient pelvic MRI to better delineate his anatomy in preparation for surgical reconstruction of his duplicated phallus. The infant has developed one urinary tract infection at home, which presented with fevers to 103°F and gross hematuria while wiping the left urethra. His urine culture was positive for *Enterobacter cloacae* and *Enterococcus faecalis*, which

was treated with cefdinir. He resumed his prophylactic amoxicillin after the urine infection was appropriately treated.

Nephrology recommended repeating renal ultrasounds after discharge to monitor bilateral hydronephrosis. His central calyceal

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dilation and ureteral dilation have improved, but the underlying

structural defects have remained unchanged.

Neurosurgery followed the neonate during and after his NICU stay due to his complex spinal anatomy. A spinal MRI at six months old showed increased conspicuity of the syrinx extending from T1–L2. The previously seen spinal, urinary, and genital structural abnormalities were noted and appeared unchanged. Surgical repair of these anomalies will be deferred until the patient is at least one year old. A repeat spinal/pelvic MRI will be repeated shortly before that time.

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An outpatient pediatric orthopedics evaluation was arranged. At nine months old, he received surgical repair of the left club foot and has recovered well so far.

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Discussion:

Diphallia is an extremely rare congenital genitourinary abnormality that is estimated to occur in 1 in 5–6 million live births (1–3). The first reported diphallia description was in 1609. Since then, there have only been about 100 cases reported in the literature, although the anomaly is likely underreported (1).

Establishing a definite cause for diphallia has been difficult, given how extremely rare and varied the presentation can be between patients. The cause of diphallia is multifactorial in most cases, with causes related to maternal exposure to teratogens, radiation, or infections; fetal injury; or an abnormal genome (3–5). While the embryologic cause of diphallia is hypothesized, several other theories have been proposed. Historic theories postulated that diphallia is due to a failure of normal fusion of the bilateral em-

bryonic anlagen, atavism, or a minor structural duplication (6). Recent studies in developmental anatomy suggest that diphallia may be due to duplication of the infra-umbilical cloacal structures, also known as caudal duplication syndrome (CDS), which leads to altered mesodermal migration and subsequent creation of two separate sets of genital structures (6–8).

A meta-analysis by Kendrick et al. evaluated 87 English-language, published case reports on diphallia presentation and associated congenital anomalies. The most common GU anomalies were single renal agenesis, horseshoe kidney, and duplicate ureters/vesicoureteral reflux. The most common GI anomalies were imperforate anus, gastrointestinal tract duplication, and anorectal malformations. The most common non-GU/GI anomalies were inguinal hernias, limb agenesis/hypotrophy, wide diastasis of the pelvic bone, hemivertebra, and meningocele/umbilical hernia/talipes equinovarus/atrial septal defect (1).

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Our case report presents a patient with a unique constellation of congenital anomalies. The patient had wholly duplicated phallic structures, a duplicated bladder, a right-sided kidney, fusion between the left and remaining right-sided kidneys, a bifid scrotum (each with two testicles), and two rectal-urethral fistulas. Other congenital anomalies included a tethered spinal cord, duplicated spinal cord at the T4 level, lipomyelomeningocele, lumbosacral spinal dysraphism, lumbosacral and pelvic dysgenesis, and a unilateral club foot/shortened leg. The unique congenital anomalies our patient presented included Tetralogy of Fallot, a right-sided aortic arch, and mirror image branching of aortic vessels.

Our patient's diphallia was likely related to CDS, given his associated genitourinary and spinal abnormalities. CDS is associated with various degrees of duplication of the genitourinary organs, lower spinal cord, and distal GI tract (9–11). Patients with CDS have sometimes been reported as having simple cardiac abnormalities like patent ductus arteriosus, atrial septal defect, and ventricular septal defect (10). However, patients with diphallia or CDS have not been reported with the more complex cardiac anomalies found in our patient. This makes our patient's cardiac abnormalities unique and an important addition to the existing literature.

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Given the wide variety of patient presentations, the treatment for diphallia CDS should be individually tailored to each patient. A detailed physical exam and imaging studies, including ultrasounds and magnetic resonance imaging, should be conducted to determine the anatomical abnormalities of the genitourinary, gastrointestinal, and musculoskeletal systems. In addition, there should be consideration for evaluation with an echocardiogram to assess for cardiac anomalies. Management often includes the discussion of surgical removal of the rudimentary/non-communicating phallus and appropriate reconstructive surgery (1–4). Patients with diphallia should receive appropriate screening and counseling for genitourinary, gastrointestinal, cardiac, extremity, and vertebral abnormalities. Minor asymptomatic abnormalities can be managed conservatively; however, additional surgical intervention may be required to address other major congenital anomalies that may be involved. Lower GI abnormalities commonly require surgical correction of anorectal malformations and duplicated gastrointestinal structures such that there is a single functioning, continent anus (5). Management of urinary tract duplications is generally more conservative, aiming to minimize urinary tract infections and maximize urinary continence (5). Even with adequate treatment, infants born with diphallia have an increased mortality rate from genitourinary and gastrointestinal infections and other complications (1, 2) The primary source of morbidity is spinal cord anomalies leading to urinary and gait disturbances in adulthood. The overall prognosis is favorable if the infant successfully survives all required surgeries. However, it is crucial to rule out chromosomal abnormalities when describing the prognosis to patients' families, as an underlying chromosomal genetic abnormality may have significant associated morbidities and mortality.

Conclusion:

Diphallia is a rare congenital developmental abnormality that presents differently with each patient. Our patient's presentation of diphallia suggests that the etiology may be related to CDS. But at this time, the pathogenesis of this congenital anomaly remains uncertain, and it may be multifactorial. Major congenital anomalies involve infra-umbilical structures of the genitourinary, gastrointestinal, musculoskeletal, and central nervous systems. Though previous reports have only described minor cardiac anomalies, our patient demonstrates that more complex cardiac abnormalities can be associated. Patients with diphallia or CDS should receive a complete cardiovascular and hemodynamic evaluation to successfully diagnose and manage all cardiac abnormalities.

Learning Points:

1. Diphallia is a rare congenital urogenital abnormality occurring at a rate of 1 in 5–6 million live births.
2. The developmental cause for diphallia is uncertain and likely multifactorial.
3. Associated anomalies include urinary, musculoskeletal, gastrointestinal, anorectal malformations, central nervous system, and cardiac anomalies.
4. Management is individualized and should be completed with a multidisciplinary team.

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Patient Consent was obtained and granted from the patient's legal guardian through our institutional written consent form. Loma Linda University Children's Hospital - Authorization and Consent to Record Audio and/or Video, Photograph, Write and Publish.

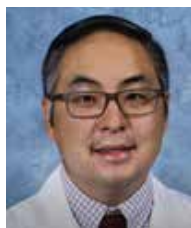
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