

Photo Quiz

Examining a Neonatal Boy With an Imperforate Anus



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Citation Mirzaee M, Khalili M, Jamee M, Mohkam M. Examining a Neonatal Boy With an Imperforate Anus. *Journal of Pediatric Nephrology*. 2022; 10(2):100-102. <https://doi.org/10.22037/jpn.v10i2.37211>

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Case Presentation

A one-day-old neonatal boy was admitted to the hospital to evaluate his imperforate anus. He was born prematurely (at 36 weeks of gestation) via normal vaginal delivery. He was the second child of non-consanguineous parents. The other sibling was healthy. The neonatal period had been uneventful.

His birth weight was 2730 g. In the physical examination, his face was not syndromic. He had an imperforated anus and bifid scrotum, but his testicles were normal size and positioned inside the scrotum. Urinary output was normal. The cardiac examination was normal. Brain sonography was normal.

In the initial laboratory test, a high serum creatinine level (0.95 mg/dL) was detected, which then decreased to 0.45.

The result of urine culture was positive for *Klebsiella* and negative following treatment.

In scrotal sonography, bilateral testicles were normal-sized; however, mild bilateral hydrocele was also observed.

In kidney and bladder sonography, the right kidney's diameter was 51×23. The left kidney was 27 mm and had multicystic dysplasia and hydronephrosis with a 15 mm Anteroposterior (AP) diameter. In addition, a cystic structure was observed in the presacrococcygeal area of the urinary bladder (diameter 27×16 mm).

Figures 1-3 show the Voiding Cystourethrogram (VCUG) and abdominopelvic Computed Tomography (CT) scans obtained at the time of presentation, respectively.

In Voiding Cystourethrogram (VCUG), the bladder showed an irregular border with the urachal diverticulum. There is no evidence of vesicoureteral reflux during filling or voiding phases.

As shown in Figures 2 and 3, the following features were observed in the CT scan:

The right kidney is 43 mm.

Left kidney hypoplasia measuring about 22 mm is observed without a significant increase in the corticomedullary phase. Cystic lesions measuring about 27 mm, 30 mm, and 40 mm are observed in presacral space, anterior to the rectum, and posterior to the bladder with displacement on mentioned organs.

The diagnosis is VACTERL (V: Vertebral anomalies, A: Anal atresia, C: Cardiovascular abnormalities, TE: Tracheoesophageal fistula, R: Renal anomalies, L: Limb defects). He has a functional kidney and neurogenic bladder due to a large cyst in the presacral space.

According to the evidence from ultrasound and CT scan, there was a suspicion of rectal duplication cyst or teratoma.

At the time of closing colostomy, the patient underwent open surgery, and the surgeon did not find the cyst in the open surgery. The diagnosis was dilated



Figure 1. Irregular border of bladder

distal pouch of the rectum. Due to a rectovesical fistula, the bladder is filled with urine and seen as a cyst.

The frequent association between Congenital Abnormalities of the Kidney and Urinary Tract (CAKUT) and bladder dysfunction shows that among the genes suspected or proved to be involved in CAKUT, some have a role in bladder embryology.

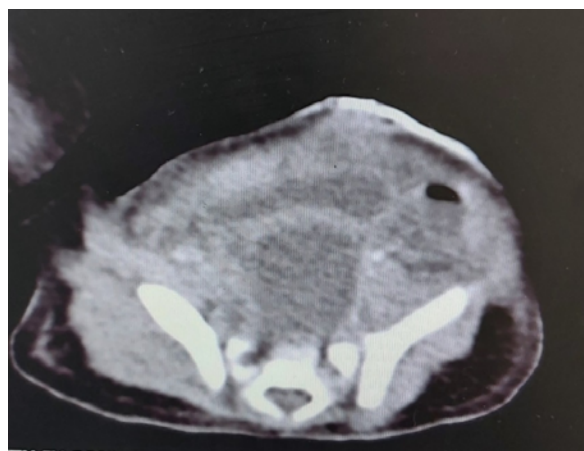
For a long time, neurogenic bladders were left untreated or treated late, with a frequent evolution to end-stage renal disease and even precocious death. A modern approach allows, in an increasing number of cases, relatively good quality of life and avoidance of end-stage renal disease [1].

Genes suggested or identified to be involved in the pathogenesis of congenital abnormalities of kidney and urinary tract (CAKUT) are assumed to play a role

in bladder embryogenesis, as bladder dysfunction is frequently observed in CAKUT patients.

In the past, the neurogenic bladder was left untreated or treated late in the majority of patients. This led to irreversible complications in affected individuals such as end-stage renal disease (ESRD) and even early death. Therefore, a modern diagnostic approach can govern a relatively good quality of life, reduce diagnosis delay and prevent the development of ESRD [1]. In this approach, the neonates are initially evaluated for urinary infection and renal insufficiency, and undergo post-voiding residual measurement (spontaneous or after the Credé maneuver) if miction can be obtained. Along with the clinical and ultrasound examinations, urodynamics can be performed by suprapubic or urethral catheterization in a timely manner for a comprehensive assessment of bladder malfunction. Application of urodynamics in the management of the neurogenic bladder also allows for early decision making regarding continuing clean intermittent catheterization (CIC)

A: A cystic lesion



B: Multicystic dysplastic kidney in left side

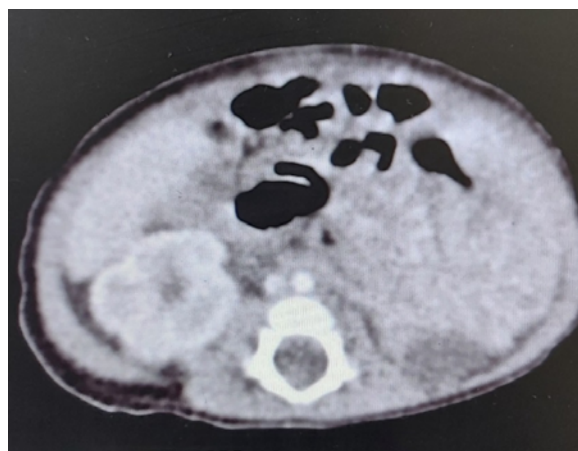


Figure 2. Cystic lesions measuring in presacral space, anterior to the rectum, and posterior to the bladder with displacement on mentioned organs.

and spontaneous miction. Anticholinergic medications can be prescribed in case of an overactive bladder. After a few months, the efficacy of the treatment may be evaluated by repeated urodynamics [2].

Ethical Considerations

Compliance with ethical guidelines

There were no ethical considerations to be considered in this research.

Funding

This project was funded by the National Agency for Strategic Research in Medical Education, Tehran, Iran. Grant No. 992150

Authors' contributions

All authors equally contributed to preparing this article.

Conflict of interest

The authors declared no conflict of interest.

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