

## Beyond borders: Prune belly syndrome unveiled in a nine-year-old Pakistani boy

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### Abstract

Prune belly syndrome (PBS) is characterised by a malformed urinary tract, undescended testes and abnormal abdominal wall musculature. No prior report of PBS is known to us from Pakistan. Most cases are diagnosed at birth, but we report the case of a nine-year-old boy who presented with the complaint of left flank pain for the past year. Upon physical examination, his abdomen was swollen with creased skin and lacked the rectus abdominis muscles. The scrotal skin had minimal wrinkling, and both the testes were undescended, along with the constriction in the external urethral meatus. This report aims to raise awareness of this syndrome for early detection and optimal treatment. It is recommended that family medicine and paediatric surgery professionals, capable of identifying and referring patients in the disease's early stages be increased, particularly in rural areas.

**Keywords:** Hydronephrosis, Megaureter, Prune-Belly Syndrome, Undescended testes.

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### Introduction

Prune belly syndrome (PBS) is a rare congenital disorder characterized by urinary tract anomalies, deficient abdominal wall musculature, and cryptorchidism.<sup>1</sup> It has an estimated prevalence of one in 40,000 live births, with 95% of cases occurring in males.<sup>2</sup> The precise etiology of PBS remains uncertain, though proposed mechanisms include mesodermal differentiation defects during the sixth to tenth week of gestation or prenatal urethral obstruction leading to abdominal wall atrophy.<sup>3</sup>

Despite its rarity, PBS poses significant diagnostic and

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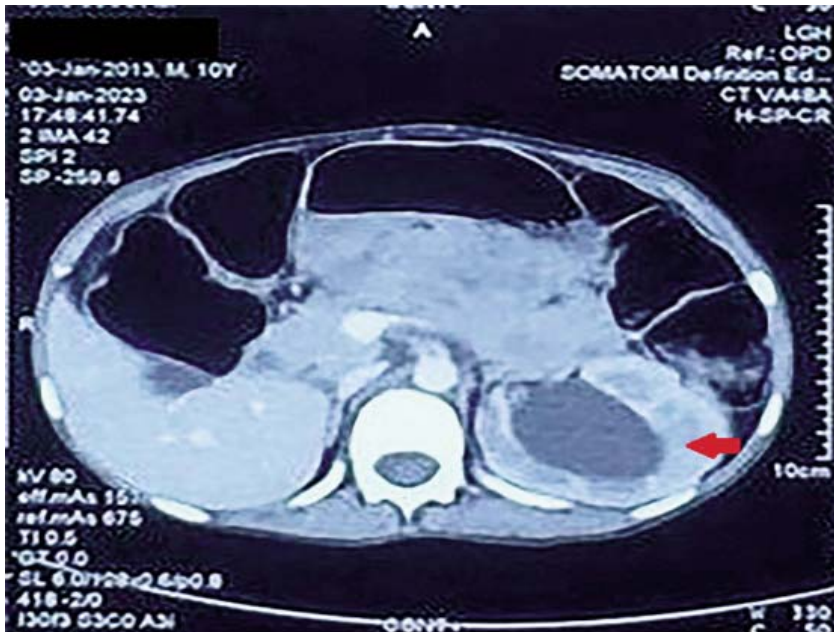
management challenges, particularly in resource-limited settings where awareness is low. Early diagnosis is essential to prevent complications such as renal failure and recurrent infections, which can severely affect the quality of life and survival. Most cases are identified during infancy or even prenatally, but late presentation, as in the current case, highlights gaps in early detection and the need for increased awareness among healthcare providers.<sup>2,3</sup>

This case report is the first documented instance of PBS in Pakistan in last twenty-three years, showcasing the condition's late presentation in a nine-year-old boy. The report aims to raise awareness among healthcare professionals, especially those in rural areas, regarding PBS's clinical features and the importance of timely intervention to prevent adverse outcomes. By presenting this case, we aim to contribute to the growing body of literature on PBS and emphasize the necessity for multidisciplinary care and enhanced diagnostic capabilities in low-resource settings.

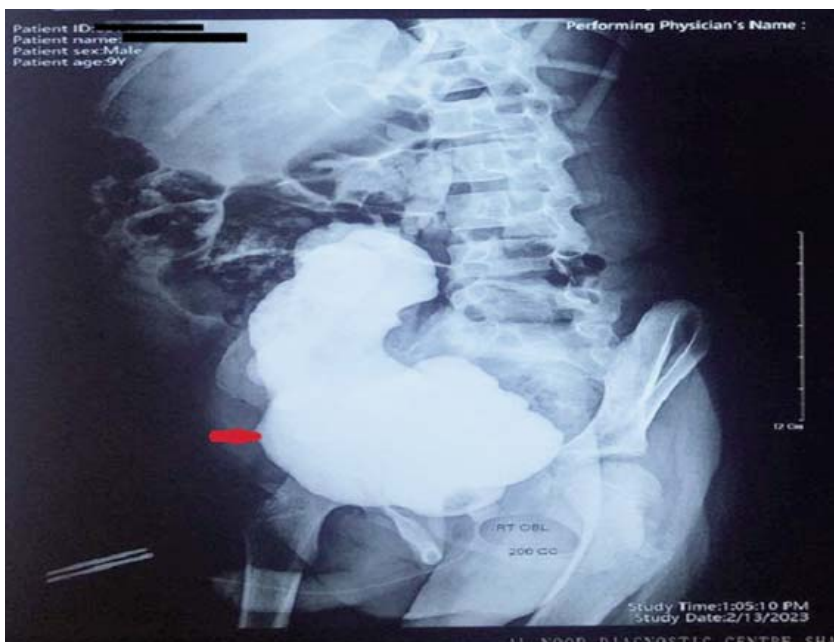
### Case Report

A nine-year-old boy from the Okara district came to the Urology Department of Pakistan Kidney and Liver Institute and Research Centre, Lahore, in January 2023, with a complaint of left flank pain since the previous year. He had been experiencing burning micturition and constipation for one month. The patient's previous medical records, as well as his family and maternal pregnancy history, were not relevant for the diagnosis of PBS. Furthermore, he is the third child in his family, and there is no history of PBS symptoms among his family members. He was otherwise healthy, with up-to-date immunisations.

On physical examination, the patient's body temperature was recorded as 38.9°C, and there was an elevation in both pulse and respiratory rates. On the other hand, the patient's blood pressure was 110/75 mmHg. His abdomen was bulging with wrinkled skin and absent rectus abdominis muscles. Scrotal skin showed little rugae. Both the testes were non-palpable, along with the narrowing of the external urethral meatus. Other systemic examinations revealed no abnormality.



**Figure-1:** CT scan of the abdomen showing left-sided marked hydronephrosis with the dilated tortuous ureter and right kidney agenesis.



**Figure-2:** Voiding cystourethrogram showing vesicoureteral reflex with a distended bladder.

Multiple red blood cells and a positive leukocyte esterase result were detected in the urine analysis. His blood urea nitrogen (BUN) level was 28 mg/dl (normal range: 7–20 mg/dl), and his creatinine level was 0.63 mg/dl (normal range: 0.5–1.2 mg/dl), both within the normal range.

The paediatric surgery team conducted an ultrasound

showing a large debris fluid collection-like structure with stretched septae in the left kidney. On a computed tomography (CT) scan, the right kidney was not well localised (Figure 1). Moderately severe cystitis was noted, with a non-developed hemi-scrotal sac and undescended testes. Micturating cystourethrogram (MCUG) showed significant residual urine in the bladder and grade 5 vesicoureteral reflux (VUR) (Figure 2).

To address undescended testes, the patient was scheduled to undergo bilateral orchidopexy. Additionally, bilateral ureteral reimplantation was planned to prevent urinary reflux and potential kidney impairment. Subsequently, a follow-up assessment was scheduled a month after the surgery to evaluate the patient's condition. The patient's parents were counselled regarding PBS and the care needed at home.

## Discussion

PBS is rare and most patients with the condition present late.<sup>4</sup> The condition is characterised by three primary abnormalities: malformations in the urinary tract, undescended testes, and deficient abdominal wall muscles.<sup>5</sup> The exact aetiology is still unknown, but two primary hypotheses regarding the pathogenesis have emerged from clinical studies, i.e. the mesodermal differentiation defect hypothesis and prenatal urethral obstruction hypothesis. According to the mesodermal differentiation defect hypothesis, abnormal development of the first lumbar myotome's derivatives during the sixth to 10th week gestation period results in incomplete or underdeveloped abdominal wall muscles and urinary tract abnormalities.<sup>6</sup> The urethral obstruction hypothesis suggests that when the

urethra is obstructed, the bladder and ureters become distended, leading to increased pressure and subsequent atrophy of the abdominal wall muscles.<sup>7</sup>

Managing individuals with PBS necessitates collaboration among a multidisciplinary team comprising a urologist,

nephrologist, paediatrician, paediatric surgeon, and anaesthesiologist. According to Woodward's classification from 1985, the patient falls into category 2 of PBS.<sup>8</sup> So, reduction cystoplasty, ureteric shortening, tapering, and reimplantation with or without abdominoplasty may be performed. It is also suggested that close monitoring be carried out, along with interventions for recurrent febrile urinary tract infections or declining renal function. Although some case series report rates of renal insufficiency as low as 7%, renal failure has been documented to occur at a rate of 20% to 30% over time.<sup>9</sup> In the present case, the patient's renal function test was within the normal range for his age, despite having a non-functioning left kidney. This could be attributed to the right kidney's compensatory function.

Most cases of PBS are identified during infancy, as reported in literature. However, in this child's case, the condition wasn't identified at birth, though he was born in a medical facility. This suggests that healthcare professionals need more awareness on the syndrome. Only a small number of PBS diagnoses, to the best of our knowledge, have been made in-utero before the newborn stage.

Multidisciplinary care is necessary for the demanding management of PBS patients. The professionals working at this centre included radiologists, urologists, paediatricians, and paediatric surgeons. Vigilance on maintaining a normal renal function is essential. Renal failure and bladder cancer have been reported to occur later in age.<sup>10</sup> Accurate diagnoses can help medical practitioners to establish appropriate procedures for follow-up care and management. This study brings to light the challenges in identifying and managing PBS during pregnancy and after delivery. It also draws attention to the detrimental effects and even fatal outcomes of the congenital disorder in low-resource environments, like Pakistan. Further studies are proposed to identify the best practices for PBS management and diagnosis in resource-constrained situations.

## Conclusion

The case report highlights the significance of intensified understanding, beforehand detection, as well as specialised care to increase outcomes, promoting increased medical facilities, especially in rural regions.

**Patient's consent:** Informed consent was taken from the patient's parents to publish this case.

**Disclaimer:** None.

**Conflict of Interest:** None.

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## AUTHORS' CONTRIBUTIONS:

**MDAS:** Concept, design, data acquisition, analysis, interpretation, final approval and agreement to be accountable for all aspects of the work.

**NBN, NZ, AUR, SM:** Concept, design, data acquisition, analysis,

interpretation and final approval.

**SI:** Drafting, revision and agreement to be accountable for all aspects of the work.