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Case Report

Prune Belly Syndrome Associated with Anorectal Malformation in a Neonate with a Normal Twin: A Case Report

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Abstract

Background: Prune Belly Syndrome (PBS), also known as Eagle-Barrett syndrome, is a rare congenital anomaly characterized by the classical triad of deficient or absent abdominal wall musculature, urinary tract abnormalities, and bilateral cryptorchidism. The syndrome predominantly affects males and is associated with a wide spectrum of genitourinary, gastrointestinal, musculoskeletal, and cardiopulmonary anomalies. The coexistence of PBS with anorectal malformation (ARM) is uncommon and represents a significant surgical and neonatal challenge. The occurrence of this rare association in one twin while the co-twin remains healthy and phenotypically normal makes the condition even more unusual and clinically significant.

Case Presentation: We present a rare case of a male neonate born as part of a twin pregnancy who was diagnosed with Prune Belly Syndrome associated with anorectal malformation. The co-twin was completely healthy with no congenital anomalies. The affected neonate presented shortly after birth with failure to pass meconium since birth, wrinkled and lax abdominal wall musculature, absent anal opening, and features suggestive of urinary tract involvement. Clinical examination and radiological investigations supported the diagnosis of PBS with ARM. The patient was admitted for stabilization and multidisciplinary management involving neonatology, pediatric surgery, and urology teams. Initial management included supportive neonatal care, fluid management, decompression, and evaluation for associated anomalies. Surgical planning was performed according to the patient's clinical status and associated congenital abnormalities. The rarity of this combination posed diagnostic and therapeutic challenges and required coordinated multidisciplinary care.

Conclusion: The association of Prune Belly Syndrome with anorectal malformation is rare and may present with significant morbidity during the neonatal period. Early diagnosis, thorough evaluation for associated anomalies, and prompt multidisciplinary intervention are essential for improving patient outcomes. Reporting such rare presentations contributes valuable information to the existing literature and increases awareness among clinicians regarding the possible coexistence of gastrointestinal and genitourinary congenital anomalies in neonates. This case also highlights the importance of careful neonatal assessment in twin pregnancies, even when the co-twin appears completely normal.

Keywords: *Prune Belly Syndrome; Eagle-Barrett Syndrome; Anorectal Malformation; Neonate; Twin Pregnancy; Congenital Anomalies; Pediatric Surgery*

Introduction

Prune belly syndrome (PBS) (Figure 1), or Eagle-Barrett syndrome, is a rare congenital disorder characterized by absent or deficient abdominal wall muscles, urinary tract anomalies, and bilateral undescended testes, predominantly affecting males (1 in 29,000 - 50,000 live births) [1].

Its pathogenesis involves early urinary tract obstruction or mesodermal developmental defects, explaining the triad and associated anomalies. Coexistence with anorectal malformation (ARM) is exceptionally rare [2]. Management is multidisciplinary, including stabilization, renal monitoring, and surgical correction [3].

This report describes a 2-day-old male neonate with PBS and high-type ARM in a discordant twin, highlighting this rare presentation and its clinical implications.

Case Presentation

A 2-day-old male neonate (Figure 1) was reported to pediatric surgical emergency in Nishtar Hospital, Multan, South Punjab. He was a product of a booked twin pregnancy, of a primigravida mother, born at approximately 32 weeks of gestation by caesarean section, with a birth weight of 1.2 kg. The parents were consanguineous and were residents of Shahjamal, south Punjab, Pakistan.

The neonate (Figure 1) presented with a lax, wrinkled abdominal wall, bilateral undescended testes, and failure to pass meconium since birth. The neonate was lethargic but responsive, not fully alert, with normal vital signs. The abdomen was soft, lax, and wrinkled (Figure 2). Bilateral testes were undescended (Figure 3) The anal opening was absent, consistent with an anorectal malformation, with no perineal fistula observed (Figure 4). Dysmorphic facial features and ear deformities were noted (Figure 5). Additionally, the neonate had bilateral clubfoot (Figure 6). Limb tone and movements were otherwise normal, and no other external anomalies were observed. The twin sibling had a normal abdominal wall, descended testes, patent anal opening, and no dysmorphic features (Figure 7).

Figure 1: *Showing a neonate with prune belly syndrome.*

Figure 2: *A lax and wrinkled abdomen.*

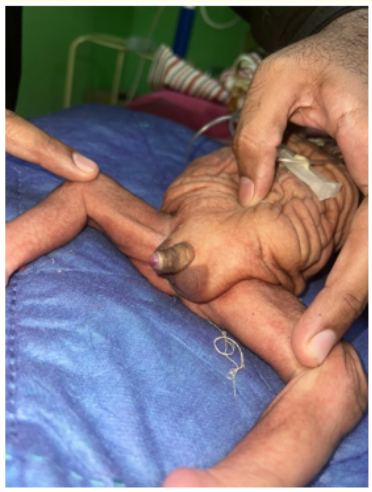

Figure 3: *Bilateral undescended testis.*

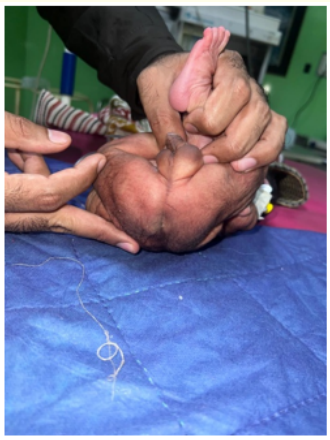

Figure 4: *Showing absent anal opening with no perineal fistula.*

Figure 5: *Showing ear and facial deformities.*

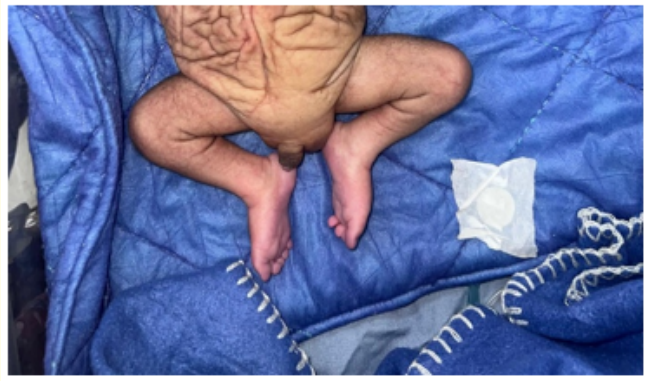

Figure 6: *Showing bilateral club foot.*

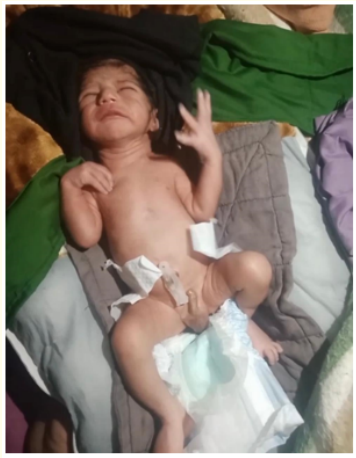

Figure 7: *Showing normal twin.*

An antenatal anomaly scan at 14.4 weeks of gestation demonstrated a large Intra-abdominal cystic lesion with bilateral moderate hydroureteronephrosis consistent with distended fetal urinary system, while the amniotic fluid volume was adequate. The pregnancy was dichorionic diamniotic, as evidenced by a thick dividing membrane and a positive lambda sign.

There was no maternal history of diabetes mellitus, hypertension, or teratogenic drug intake. At birth, the neonate did not cry immediately and required resuscitation, while APGAR scores were unavailable. The twin sibling was clinically normal, with no detectable congenital anomalies.

Upon admission, the neonate was stabilized by the pediatric surgery team. An intravenous (IV) line was secured, and supplemental oxygen was provided. Initial resuscitation included approximately 60 mL of 10% dextrose water, administered to correct hydration and support perfusion. The neonate was closely monitored for vital signs, urine output, and clinical response prior to definitive surgical management.

Postnatal neonatal ultrasound demonstrated bilateral hydronephrosis (Figure 8) with a lax abdominal wall. Despite the structural abnormalities, the neonate was passing urine adequately, indicating preserved renal function. An x-ray abdomen cross table lateral view confirmed a high-variety anorectal malformation (Figure 9).

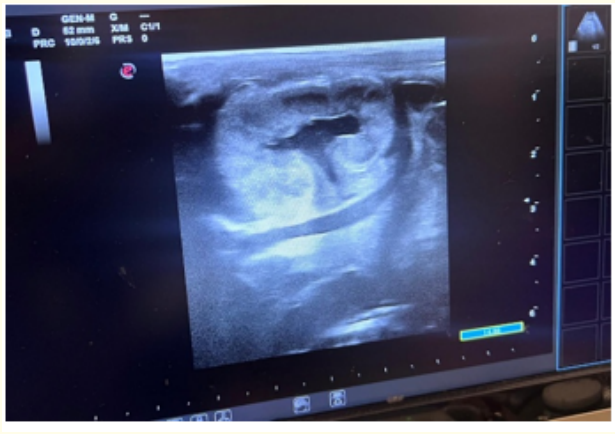

Figure 8: *Showing ultrasound with hydronephrosis.*

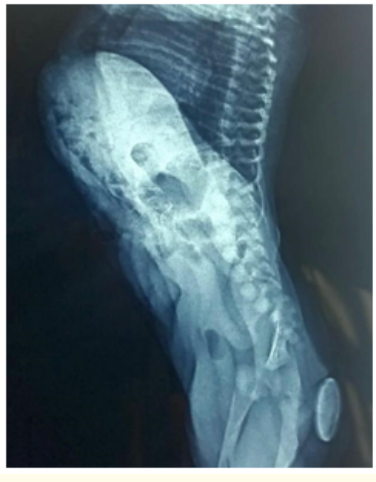

Figure 9: *Showing high variety anorectal malformation on x-ray abdomen cross table lateral view.*

Laboratory investigations revealed a normal complete blood count, but electrolyte disturbances with hyperkalemia (6.85 mmol/L) and hypernatremia (150 mmol/L) were noted. Renal function was impaired, with rum creatinine 1.61 mg/dL and urea 66.9 mg/dL. Liver function tests were deranged, showing AST 42.6 IU/L, ALT 7.85 IU/L, and ALP 99.4 IU/L.

	Values
Hb	11g/dl
TLC	6.51/mm^3
Platelets	266×10^3/ul
S/K^+	6.85 mEq/L
S/Na^+	150 mmol/L
S/ Cl^-	118.9 mEq/L
AST	42.6 U/L
ALT	7.85 U/L
Alkaline Phosphatase	99.4
Total bilirubin	5.59 mg/dl
Serum urea	66.9 mg/dl
Serum creatinine	1.61

Table

Differential diagnoses included Megacystis-Microcolon-Intestinal Hypoperistalsis Syndrome (MMIHS), posterior urethral valves (PUV), and isolated urinary tract anomalies. MMIHS typically presents with severe intestinal hypoperistalsis and lacks the abdominal wall laxity and cryptorchidism seen in PBS. PUV presents with hydronephrosis but usually has a normal abdominal wall and anal opening. Other syndromes, such as Beckwith-Wiedemann, involve macrosomia or organomegaly, which were absent. The presence of the classic PBS triad along with anorectal malformation confirmed the diagnosis.

A sigmoid colostomy was performed (Figure 10), and meconium plugs were gently evacuated to establish bowel patency. Care was taken to preserve surrounding structures and ensure proper stoma placement.

Patient was discharged on second post op day with proper stoma care guidance and further follow up.

Figure 10: *Showing sigmoid colostomy of neonate with prune belly syndrome.*

Postoperatively, the neonate was monitored for vital signs, urine output, and stoma function. Feeding was initiated gradually once tolerated, and the stoma remained functional with adequate bowel output. The neonate's condition remained stable, and plans were made for staged definitive repair of the anorectal malformation once clinically optimized.

Discussion

Prune belly syndrome (PBS), also known as Eagle-Barrett syndrome, is a rare congenital anomaly defined by a classic triad of deficient abdominal wall musculature, urinary tract malformations, and bilateral cryptorchidism. The condition has an estimated incidence of approximately 1 in 30,000-50,000 live births, with over 95% of affected individuals being male [4].

Most cases are recognized in the neonatal period due to the characteristic physical findings or antenatal ultrasound abnormalities such as urinary tract dilatation [5].

In our patient, the antenatal scan demonstrated a large intra-abdominal cystic lesion with bilateral hydronephrosis, a finding consistent with distended urinary structures that typify the underlying pathophysiology of PBS. However, unlike typical PBS presentations where urinary stream abnormalities are prominent, this neonate was passing urine adequately postnatally, despite hydronephrosis, suggesting variable functional impact.

While musculoskeletal abnormalities such as talipes (clubfoot) and facial dysmorphism may be seen in a proportion of PBS cases, the coexistence of a high anorectal malformation (ARM) is exceedingly rare. Gastrointestinal anomalies are reported in a minority of patients, and ARM in the setting of PBS has been described only sparingly in the literature [6].

Earlier case reports have included associations with high ARM and other significant complications, underscoring the complexity and heterogeneity of PBS presentations.

Investigations in PBS focus on postnatal imaging to delineate the urinary tract and rule out associated anomalies. In this case, neonatal ultrasound confirmed bilateral hydronephrosis, while invertogram demonstrated a high-type ARM, consistent with clinical examination. Laboratory tests revealed electrolyte disturbances and renal function impairment, a pattern often seen in significant urinary tract involvement.

Management of PBS is individualized and often multidisciplinary, involving initial stabilization with fluids and supportive care, followed by surgical correction of obstructive or functional anomalies. In our case, after stabilization including intravenous fluid and supplemental oxygen, a sigmoid colostomy was performed for ARM, with evacuation of meconium plugs and establishment of a patent stoma. Definitive repair of the malformation will be pursued as the child's condition permits.

The concurrence of PBS with ARM, dysmorphic features" and clubfoot in a discordant twin pregnancy underscores both the rarity and variability of this condition and suggests a sporadic developmental defect rather than a simple genetic etiology. Reporting such unusual presentations enriches the existing literature and heightens awareness of the broad spectrum of anomalies that may accompany PBS.

Conclusion

Prune belly syndrome is a rare congenital disorder with a wide spectrum of clinical presentations. The coexistence of prune belly syndrome with a high-type anorectal malformation in a discordant twin neonate represents an exceptionally uncommon association, suggesting a sporadic developmental etiology rather than a hereditary cause. This case highlights the importance of thorough antenatal evaluation, early postnatal recognition, and timely surgical intervention in optimizing outcomes. Reporting such rare associations contributes to a better understanding of the heterogeneity of prune belly syndrome and underscores the need for individualized, multidisciplinary management in affected neonates.

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