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Prune Belly Syndrome, the Unusual Presentation of the Rare Pathology

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Abstract

Prune-belly syndrome (PBS) is a rare congenital disorder that mainly affects males and is manifested by a triad of weak or deficient abdominal wall muscles, urinary tract malformation, and cryptorchidism. It has an estimated incidence of 2 - 3 per 100,000 live births. Although it has been reported to almost always affect males, there have been a few reported cases of females affected with PBS in the literature [1].

We hereby report three cases of PBS delivered at King Abdullah University Hospital (KAUH), a tertiary academic hospital in North of Jordan. Each of these cases has something peculiar that makes this series worth reporting. We also conducted a thorough literature review about PBS and included a quick review about presentation, management and suggestions.

Keywords: *Prune Belly Syndrome; Neonates; In Utero; Nephrostomy*

Introduction

Prune-belly syndrome (PBS) is a rare congenital disorder that mainly affects males and is manifested by a triad of weak or deficient abdominal wall muscles, urinary tract malformation, and cryptorchidism.

Cases

Case no. 1

A 34-week late preterm female patient born by emergency Cesarean section due to non-reassuring fetal heart rate after the mother presented with preterm labor pain and spontaneous rupture of membranes. Birth weight was 2500 grams and APGAR score 1,5 and 7 at 1,5 and 10 minutes respectively. She was diagnosed antenatally to have megacystis and so she underwent in utero vesicocentesis multiple times. Following delivery, she was noticed to have wrinkled abdominal wall with normal female external genital exam. Ultrasound of the renal system showed mild bilateral hydronephrosis and a nearly empty urinary bladder with significant ascites and no other renal anomalies, this finding raised the suspicion of urinary bladder injury following the intrauterine intervention. Her kidney function tests and electrolytes were within acceptable ranges. She was treated conservatively, ascitic fluid was drained via peritoniocentesis and she

was placed on antibiotics. Subsequently, the infant started to urinate, repeated renal ultrasound showed improvement in hydronephrosis and resolution of ascites. Her systemic evaluation revealed ASD and PDA by cardiac echocardiogram without other systemic involvement.

Case no. 2

A 33-week preterm infant of diabetic mother male born by emergency Cesarean section secondary to polyhydramnios and large-for-gestational age. Birth weight was 2580 grams and APGAR score 1,4 and 8 at 1,5 and 10 minutes respectively He was diagnosed antenatally to have bilateral hydronephrosis. After birth, the baby was intubated and placed on mechanical ventilation for respiratory distress. On exam, he was noticed to have redundant abdominal wall with bilateral undescended testes and left sided inguinal hernia. Postnatal ultrasound revealed bilateral hydronephrosis with tortuous hydroureters and non-distended bladder. Systemic evaluation revealed ASD and PDA by cardiac echocardiogram and early periventricular leukomalacia changes by head US. Due to spontaneous urination and normal kidney function tests, the infant was started on prophylactic antibiotics with no surgical interventions performed. At the age of 8 days, the infant exhibited symptoms of sepsis in the form of lethargy and temperature instability, full septic work up was done and antibiotics started. Blood and urine cultures grew *Candida* species, so Amphotericin B was added. Renal US was repeated and showed fungal balls in the urinary bladder and renal pelvicalyceal system. Due to persistent candiduria and no improvement in the US findings of fungal balls, the infant received Amphotericin B infusion transurethrally and underwent emergency nephrostomies through which Amphotericin B was also instilled daily for 7 days. Urine culture was cleared of *Candida* and follow up ultrasound showed complete disappearance of fungal balls.

Case no. 3

A 33-week preterm male, part of spontaneous triplet gestation, delivered by Cesarean section with a birth weight of 2080 grams and APGAR score 6 and 9 at 1 and 5 minutes respectively Antenatally, this baby was diagnosed to have bilateral hydronephrosis. On exam, he was found to have distended wrinkled abdomen with bilateral cryptorchidism. Postnatal US showed bilateral hydronephrosis with bilateral tortuous hydroureters and overdistended bladder. MCUG showed dilated proximal urethra suggestive for posterior urethral valve (PUV). His creatinine level peaked at 100. He was managed by transurethral foley's catheter with much improvement of the hydroureteronephrosis on subsequent US. He was discharged home on prophylactic antibiotics and given an appointment with the urologist for elective repair of PUV. Systemic evaluation revealed ASD and PDA by cardiac echocardiogram with no other system involvement. The other triplets were healthy.

Discussion

PBS, also known as Eagle Barett or Triad syndrome, is a very rare congenital syndrome with an incidence of nearly 1/100,000 with more than 95% of cases being males. This syndrome was reported for the first time in the 1800's [2]. Still, the underlying etiology remains unknown although some studies have suggested genetic inheritance [3].

With the advance in fetal medicine care, most cases can be diagnosed antenatally by US. In our report, all three cases were suspected to have PBS by antenatal US.

The majority of PBS patients have associated anomalies affecting other body organs like the heart, brain, gastrointestinal tract (GI) and musculoskeletal system (MSS) [4]. All three cases in our report had associated PDA and ASD following cardiac evaluation which is consistent with what has been reported in multiple case series [4,5]. None of our cases had any associated GI or orthopedic abnormalities although these have been reported to affect 20 - 50% of cases [6,7].

Our first reported case in this series was peculiar in that it affected a female with no external genital anomalies who underwent successful in utero vesicoamniotic drainage. There are few reported cases of females with PBS associated with genital or cloacal anomalies [8]. Perez., *et al*. reported a case of female PBS who underwent an in utero intervention with vesicoamniotic shunt at 17 weeks gestation followed by vesicostomy after birth for severe bladder outlet obstruction with excellent outcome [9].

Our second reported case was chosen due to the unusual complication of Candidal UTI and fungal balls with the excellent response to local instillation of Amphotericin B transurethral and via nephrostomy tubes. This management of fungal UTI has been reported in patients without PBS with favorable response [10].

Our third case was the first reported case in literature about a PBS affecting one baby of triplet gestation. Balaji et al reported a concordant PBS among monozygotic twins whereas R Patel reported a case of PBS among a non-identical twin [11]. Their findings might support the genetic theory behind the occurrence of this syndrome but no definitive genetic abnormality has been linked to this syndrome so far.

In view of the wide range of severity, from intrauterine fetal demise to near normal life expectancy, management needs to be individualized. Despite the advance in medical and surgical interventions, the mortality rate remains high due to pulmonary hypoplasia and end stage renal disease [12]. For survivors, reconstruction of the urinary tract abnormalities together with abdominoplasty and orchiopexy are the most common surgical interventions to improve the quality of life [13].

Conclusion

In conclusion, PBS is diagnosed based on clinical features. Although it is very rare, antenatal diagnosis can be achieved using good ultrasonographic techniques with potential intrauterine interventions to improve outcome. Thorough work up should be established after birth to confirm diagnosis and screen for associated systemic anomalies. Early surgical team approach is expected to improve the quality of life of affected patients and should be introduced soon after birth.

Ethical Standards

Parents of infants gave a full consent to have those cases published.

Funding Resource

NA

Author Declaration

Both authors declare no conflict of interest.

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