ISSN NO. 2320-5407

Journal homepage: http://www.journalijar.com

INTERNATIONAL JOURNAL OF ADVANCED RESEARCH

RESEARCH ARTICLE

Prune belly syndrome with urethral hypospadiasis, hypoplasia, imperforate anus, recto - vesical fistula and bilateral CTEV: A case report

Altaf Ahmad Bhat (MBBS, DCH),Tania chakarborty(MBBS), Pankaj kumar(MBBS, DCH), Shambhavi Tewary(MBBS).

From Department of Pediatric, Peerless Hospital and BK Roy Research Center, 360 Panchasayar, Kolkata 700094

Manuscript Info

Manuscript History:

Received: 18 May 2014
Final Accepted: 20 June 2014
Published Online: July 2014

Key words:

Corresponding Author

Altaf Ahmad Bhat

Abstract

Association between Prune belly syndrome (PBS) with urethral, hypospadiasis, imperforate anus and recto-vesical fistula is an unusual condition. It is usually fatal unless there is a communication between the fetal bladder and the amniotic sac. We report a case of PBS with urethral hypospadiasis and hypoplasia, imperforate anus congenital recto-vesical fistula and b/l CTEV deformity in a male neonate. Patient had severe respiratory distress syndrome at birth (HMD) received prolonged mechanical ventilation and surfactant. Patient was passing meconium and urine through ventrally located urethral orifice. Patient survived early neonatal period, but died due to severe sepsis and multiorgan failure on 20th day of life.

Introduction

Prune belly syndrome (PBS) is a rare genetic disease affecting about 1 in 40,000 births and almost 97% of those affected are male.[1] PBS is characterized by deficient abdominal wall musculature, hypotonia, urinary tract dilatation and bilateral intra-abdominal testes.[1,2,3] Although the etiology of PBS has not yet been well defined, some hypothesis have been suggested such as distal obstructive uropathy and mesodermal defect of the abdominal wall and the urinary tract.[1,2,3]

Urethral hypoplasia is one of the associated urologic abnormalities in PBS and present in around 18% of cases.[4] In this condition, prognosis will be very poor and it may lead to death unless there is an associated patent urachus or recto-vesical fistula, some case are reported with vesico-cutaneous fistula. We report a case of PBS with urethral hypospadiasis associated with recto - vesico fistula and imperforate anus.

CASE REPORT

A 32 weeks gestational age male neonate Second of twin delivered through LSCS was referred to our NICU for prematurity severe respiratory distress with grunt, downes score less than 6 and with a history of prenatal bilateral hydrouretronephrosis, oligohydramnios. Clinical examination revealed typical appearance of PBS with no dysmorphic facies. Abdominal examination showed a bilateral palpable kidneys and distended urinary bladder . Abdominal wall was lax with loose skin folds. Bilateral CTEV present. Genital examination showed a phallus length of 2.5 cm with hypospadiasis draining meconium and urine from same orifice, testis were bilaterally non-palpable, empty scrotum, bilateral undescended testes and urine stained black with meconium dribbling from a ventral hypoplastic urethral orifice(Fig 1a & Fig 1b). Trial of insertion of urethral catheter was unsuccessful. Laboratory investigation showed creatinine of 2.8mg/dl

Ultrasound revealed grade IV to V bilateral hydronephrosis (Fig 2)

Voiding cystourethrogram (VCUG) showed bilateral high grade vesico-ureteric reflux (VUR) with dilated posterior urethra and hypoplastic anterior urethra

The chest X Ray showed horizontal and crowding of ribs with complete white out lung fields (RDS Syndrome)

DISCUSSION

Prune belly syndrome is a broad spectrum of anatomic defects and different levels of severity. Severe cases are most probably stillborn due to congenital lung and kidney hypoplasia from oligohydramnios.[3,4] However, survivors will have a varying degree of renal affection throughout their lives. Our patient had the imperforate anus and with a communication between rectum and urinary badder. Being premature with significant pulmonary hypoplasia and grossly hydronephrotic kidneys, the baby was fighting with potential lethal anatomic abnormalities, and so was our team dealing with a sick baby with rare clinical presentation.

Although the anterior urethra of the PBS child is usually normal, a spectrum of urethral maldevelopment of the anterior urethra has been reported, urethral atresia (microurethra) and megalourethra being the most common.[5,6]

Unless it is associated with a patent urachus or a spontaneous vesico-cutaneous fistula, urethral atresia is often lethal[4,7]. Our case had a communication between urinary system and gastrointestinal tract . It has been postulated that urethral atresia or microurethra occurs because the urethra is unused rather than malformed. Spontaneous bladder rupture with fistula formation has been reported by Reinberg *et al*. in 1993[4]

There are only few reports about the association between PBS and urethral hypoplasia in the literature.[4,7,8] Reinberg and colleagues[4] reported 6 patients with PBS and urethral atresia (3 boys and 3 girls). Three of them died from pulmonary hypoplasia and three survived. During follow up, two out of three survivors had renal insufficiency while only one had normal renal function.

Gonzalez *et al*., 2001[7] documented 5 patients who had PBS with urethral atresia. VCUG identified moderate to high grade VUR in all patients with a nadir serum creatinine at age 1 year of 1.3 mg/dl (range 0.5 to 2.1). All patients were treated initially with vesicostomy and thereafter with multiple urological procedures. Renal failure developed in 4 patients (80%) before the age of 10 years. He concluded that although urethral atresia is incompatible with life, prenatal decompression allows survival and in some cases may even lead to normal bladder and renal function. The majority of their cases required some form of supravesical diversion and reconstruction of anterior abdominal wall in a series of step wise operative procedures .

Urinary diversion may be necessary as a temporary measure in children with acute renal failure, urinary sepsis, or bladder outlet obstruction from urethral atresia with limited patency of the urachus. When temporary urinary diversion is indicated, a cutaneous vesicostomy is the procedure of choice. This is best done by the Blocksom technique as described by Duckett and colleagues (1974).[9].

Treatment of urethral hypoplasia in PBS includes gradual progressive dilation using soft catheter with insertion of stent, urethrotomy and open reconstruction.[10,11,12] However, conflicting data have been reported Passerini-Glazel and colleagues (1988)[10] reported on progressive gentle urethral dilation with good success. This technique may be used *in situ* or an antegrade and retrograde fashion in cases in which a vesicostomy has been performed. As reported by Reinberg and colleagues (1993),[4] however, this technique is not uniformly successful and may require a more formal urethroplasty with skin flaps or grafts, or both. Kajbafzadeh *et al*., 2010[8] reported 7 infants with PBS and urethral hypoplasia presented either with open urachus or surgically created urinary diversion. They documented the success of urethral hydro distension for the management of urethral hypoplasia. Follow-up imaging studies showed a significant improvement in all patients except one.

Our baby survived early neonatal period but died due to late onset sepsis and multiple organ failure, instead we tried our best but these babies have to undergo series of stepwise operative procedures and most of them who survive suffer renal insufficiency and end stage renal disease.

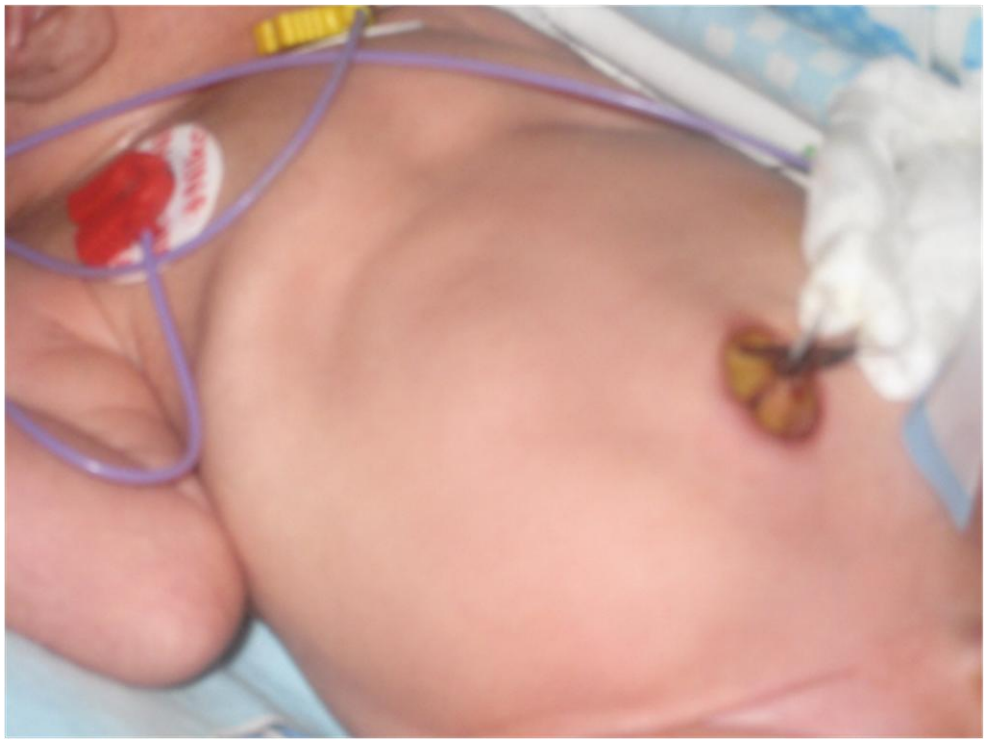

Figure 1a

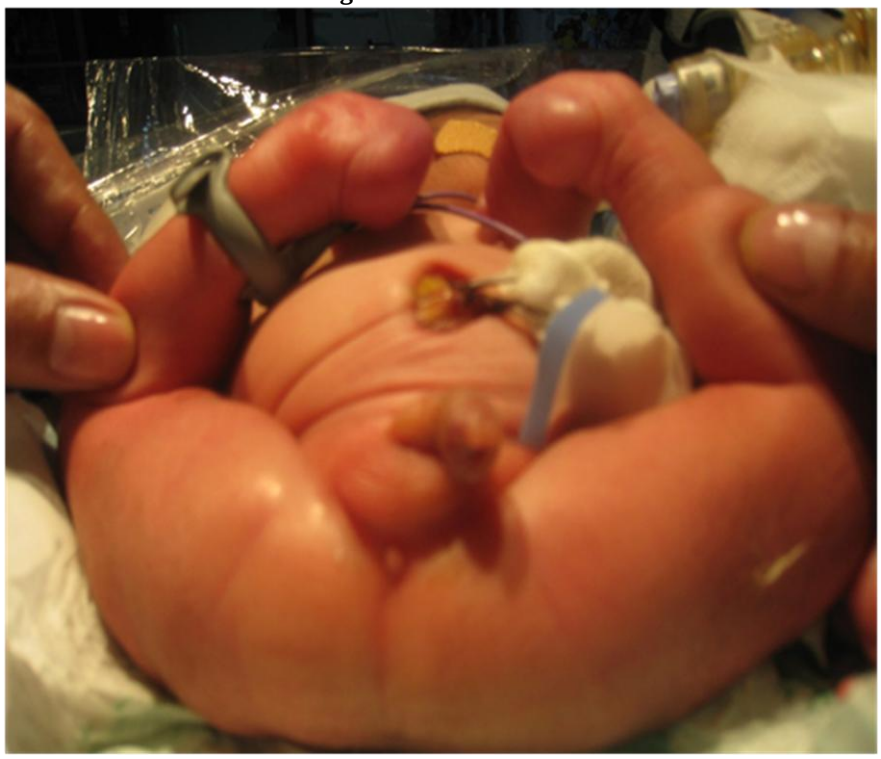

Figure 1b.

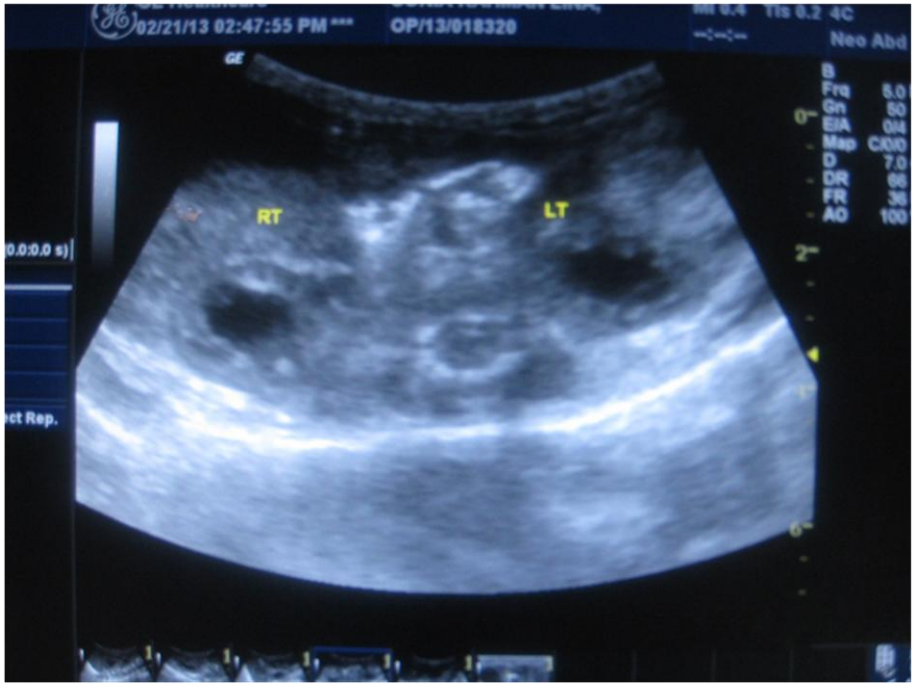

Figure 2.

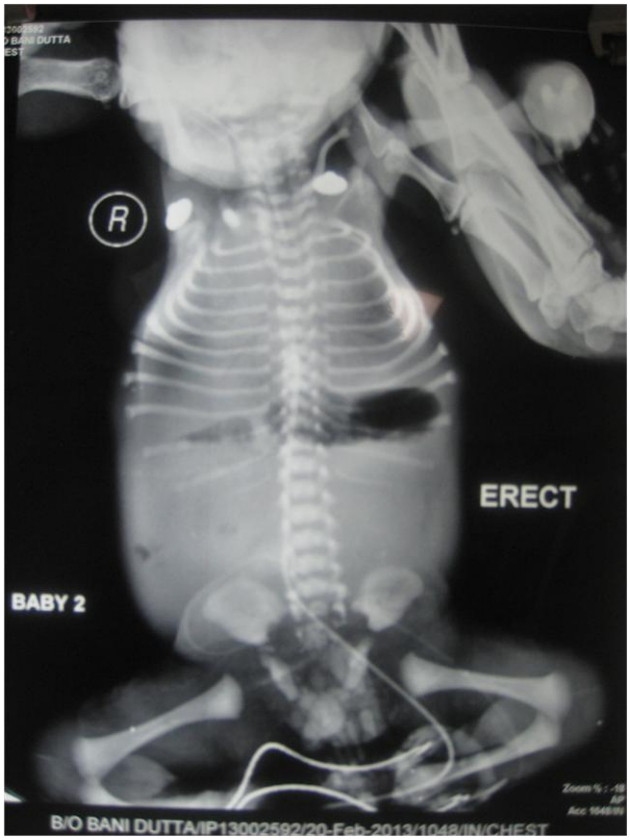

Figure 3

Source of Support: Nil

Conflict of Interest: None.

REFERENCES

1. Hubinois P, Valayer J, Cendron J. A series of 34 cases of prune belly syndrome in children. Sem Hop. 1983;59:2769–77.

2. Stephens FD, Gupta D. Pathogenesis of the prune belly syndrome. J Urol. 1994;152:2328–31.

3. Woodard JR, Smith EA. Prune belly syndrome. In: Walsh PC, Retik AB, Vaughan ED Jr, Wind AJ, editors. Campbell's urology. 7th ed. Vol. 4. Philadelphia: WB Saunders; 1998. pp. 1917–38.

4. Reinberg Y, Chelimsky G, Gonzalez R. Urethral atresia and the prune belly syndrome. Report of 6 cases. Br J Urol. 1993;72:112–4.

5. Kroovand RL, Al-Ansari RM, Perlmutter AD. Urethral and genital malformations in prune belly syndrome. J Urol. 1982;127:94–6.

6. Perrotin F, Ayeva-Derman M, Lardy H, Cloarec S, Lansac J, Body G. Prenatal diagnosis and postnatal outcome of congenital megalourethra. Report of two cases. Fetal Diagn Ther. 2001;16:123–8.

7. González R, De Filippo R, Jednak R, Barthold JS. Urethral atresia: Long-term outcome in 6 children who survived the neonatal period. J Urol. 2001;165:2241-4.

8. Kajbafzadeh AM, Rasouli MR, Dianat S, Nezami BG, Mahboubi AH, Sina A. Urethral hydrodistension for management of urethral hypoplasia in prune belly syndrome: Long-term results. J Pediatr Surg. 2010;45:2217–21.

9. Duckett JW., Jr Cutaneous vesicostomy in childhood. The Blocksom technique. Urol Clin North Am. 1974;1:485–92.

10. Passerini-Glazel G, Araguna F, Chiozza L, Artibani W, Rabinowitz R, Firlit CF. The P.A.D.U.A. (progressive augmentation by dilating the urethra anterior) procedure for the treatment of severe urethral hypoplasia. J Urol. 1988;140:1247–9.

11. Al-Bassam A, Sheikh MA, Al-Smayer S, Al-Boukai A, Al-Damegh S. Congenital H-type anourethral fistula with severe urethral hypoplasia: Case report and review of the literature. J Pediatr Surg. 1998;33:1550–3.

12. Scherz HC, Kaplan GW. Etiology, diagnosis, and management of urethral strictures in children. Urol Clin North Am. 1990;17:389–94.