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Prune Belly Syndrome: Two Case Series.

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Abstract:*Prune Belly syndrome (PBS) is a rare congenital anomaly and is almost exclusive to males. We are reporting 2 cases of PBS. The first case of 8 days old baby born to a mother (29 years) who has history of such child previously but unfortunately that baby didn't survive and died at 1.5 years of age. On physical examination child had wrinkled skin over abdomen showing signs of deficient abdominal wall, undescended bilateral testis and palpable kidney. Ultrasound examination of the abdomen revealed bilateral gross hydrouerteronephrosis and patent urachus. The second case is of 18 months old boy born out of consanguineous marriage with deficient abdominal wall, palpable kidney and bilateral undescended testis. In this case USG was suggestive of left hydroureteronephrosis. Micturatingcystourethrogram(MCU) was showing bilateral VUR.The findings were suggestive of prune belly syndrome in both cases and hence further work up was done.Early diagnosis is important for complete work up and the optimal management.*

Keywords: *Bilateral undescended testis, hydronephrosis, Prune Belly syndrome, patent urachus.*

I. Introduction

Prune-belly syndrome (PBS) is a rare congenital syndrome with an estimated incidence of 1 in 35,000 to 1 in 50,000 live births.It occurs almost exclusively in males (>95%),[1]and is seen more frequently in twin pregnancies.It is characterized by deficient development of abdominal muscles that causes the skin of the abdomen to wrinkle like a prune, bilateral cryptorchidism, abnormalities of the urinary tract such as bilateral gross hydronephrosis and megaureter.Though the cause is unknown but one theory suggests that there is a mesenchymal insult to the fetus at ~6 weeks gestation secondary to a migrational defect of the lateral mesoblastresultingin deficient abdominal muscular development. A second theory suggests that the problem may be secondary to chronic intrauterine abdominal distention with subsequent pressure atrophy of abdominal muscles[3]The prognosis is generally poor with high infant death. We report a case of PBS with genetic association.

II. Case Report

A 29 year old female came in OPD with complain of abnormal looking child with wrinkles over the skin. This was antenatally booked case and child was born out of non-consanguineous marriage. There was no history of drug allergy, fever or allergy to mother. On physical examination there was wrinkling of skin over abdomen with deficiency of abdominal wall musculature, palpable kidney, scoliosis, clubfoot, bilateral undescended testis and urine was coming out of umblical region suggestive of patent urachus. [Figure 1.] The ultrasonography showed bilateral hydroureteronephrosis and patent urachus.The CECT abdomen and urography reported abdominal wall defect, urinary bladder with marked circumferential wall thickening. There was marked dilatation of bilateral pelvicalyceal system with dilated and tortuous ureters of both sides.[Figure2]The echocardiography study was normal in this child. The child was catheterized and was given antenatal care.

The second case was a male child of 18months when reported in Pediatric surgery OPD with complaints of distended abdomen, dribbling of urine and bilateral undescended testis (UDT). [Figure 3]On physical examination left sided kidney was palpable with bilateral UDT. On ultrasonography left sided dilated pelvicalyceal system and ureter was seen. The abdominal x ray was showing bulge on right side due to abdominal muscle weakness.[Figure 4]While MCU revealed bilateral VUR and posterior urethral valve(PUV).[Figure 5]

III. Discussion

Although PBS (also known as Eagle-Barrett syndrome) is characterized by the classical triad of urinary tract anomalies, deficient abdominal musculature, and bilateral cryptorchidism, association with other anomalies including musculoskeletal, cardiovascular, pulmonary and genital malformations have been reported in the literature.[2] Haeri*et al*. in 2010[4] have reported the association of PBS with an apparently *de novo* 1.3 megabase interstitial 17q12 microdeletion that includes the hepatocyte nuclear factor-1-beta gene at 17q12, and the authors suggested that haploinsufficiency of hepatocyte nuclear factor-1-beta may be causally related to the

production of the PBS phenotype through a mechanism of prostatic and ureteral hypoplasia that results in severe obstructive uropathy with urinary tract and abdominal distension.Although the primary molecular defect underlying PBS remains unclear, clinical studies have given rise to two main pathogenic hypotheses; these are the mesodermal defect hypothesis and the urethral obstruction malformation complex hypothesis. According to mesodermal defect hypothesis [5] aberrant development of the derivatives of the first lumbar myotome between 6 and 10 weeks of gestation leads to a patchy muscular deficiency or hypoplasia of the abdominal wall as well as to urinary tract abnormalities. The urethral obstruction malformation complex hypothesis [6] proposes that pressure atrophy of the abdominal wall muscles occurs when urethral obstruction leads to massive distension of the bladder and ureters. Bladder distension would also interfere with descent of the testes and thus be responsible for the bilateral cryptorchidism.
Our patient comes under category 3 PBS as described by Woodard in 1985.[Table1]

Table1. Classification of PBS (Woodard 1985)

Table 1

Classification of PBS (Woodard 1985)

Category	Characteristics
I	Renal dysplasia
	Oligohydramnios
	Pulmonary hypoplasia
	Potter's facies
	Urethral atresia
II	Full triad features
	Minimal or unilateral renal dysplasia
	No pulmonary hypoplasia
	May progress to renal failure
III	Incomplete or mild triad features
	Mild to moderate uropathy
	No renal dysplasia
	Stable renal function

Our patient was managed with teamwork from various departments like neonatology, nephrology, urology, orthopedics and others. The child can be managed with MCU to rule out bladder outlet obstruction (BOO) and vesicoureteric reflex(VUR). In cases with BOO, suprapubic catheterization is performed to decompress urinary system. While chest X ray can give us the idea about pneumothorax, pulmonary hypoplasia and pneumomediastinum.The first case was of category 3 and second case of category 2. In our patients if renal insufficiency develops then we can do ureteric shortening, tapering and reimplantation with or without abdominoplasty (Monfort technique). Careful monitoring is required to avoid any febrile episodes of UTI. In cases of repeated episodes and renal deterioration active intervention is required.

Diao*et al*. have reported renal failure to be the main cause of death in PBS.[7] Serum creatinine levels is a useful predictor of long term renal function and levels less than 60 µmol/l indicate good prognosis. Although the condition is usually incompatible with life due to visceral abnormalities especially that of renal function, there are cases who survived into adult life after urinary tract repair and abdominal reconstruction surgery.[8] When diagnosed in the antenatal period by ultrasonography intrauterine therapeutic option including in utero placement of a vesicouterine shunt can be done to prevent the development of PBS, as reported by Leenerse*t al*.[9]

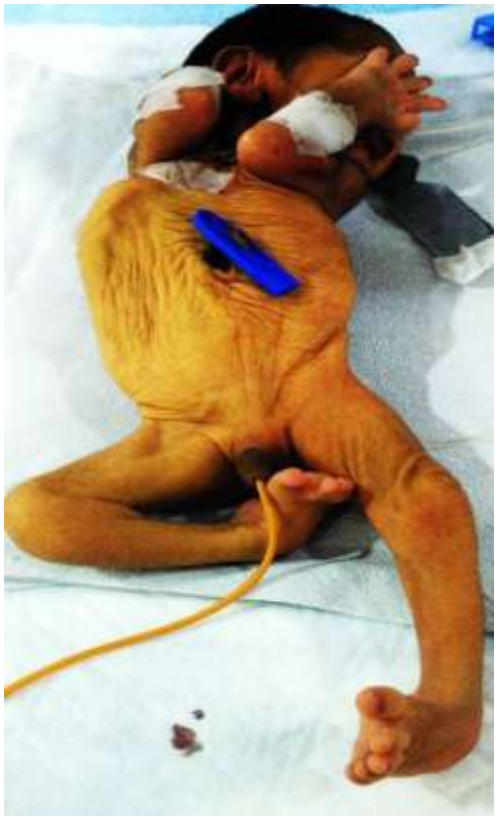

Fig 1. First case of PBS showing abdominal muscle weakness, spinal and foot deformity

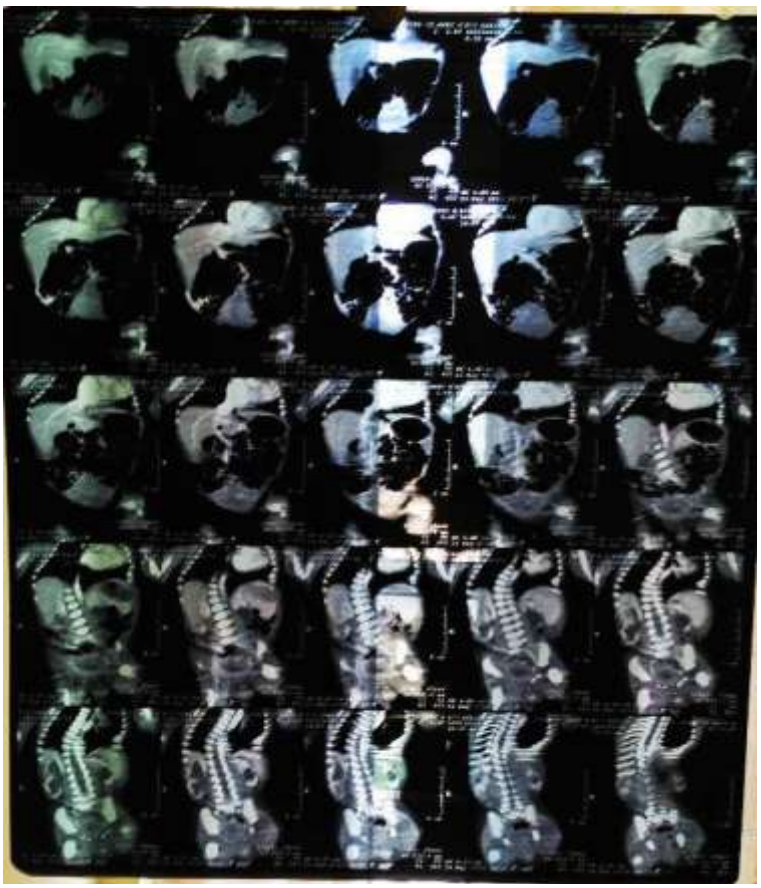

Fig 2: Showing spinal deformity along with Bilateral Hydroureteronephrosis.

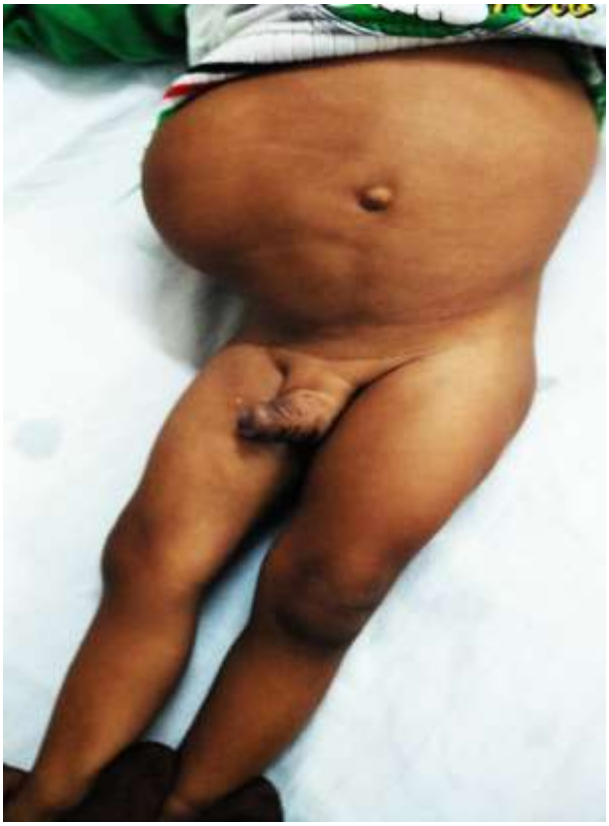

Fig 3. Second case of PBS with palpable left kidney, deficient abdominal muscles and bilateral undescended testis.

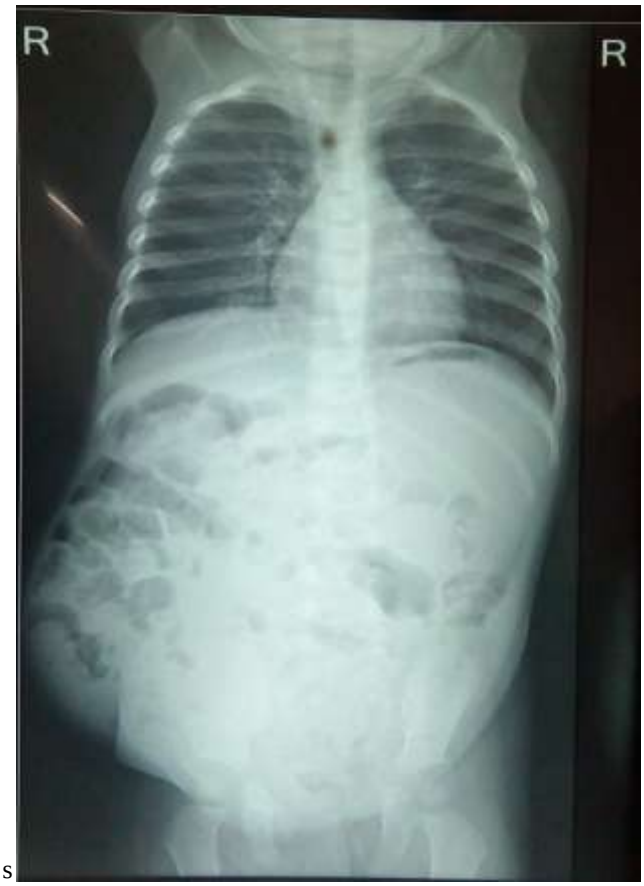

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Fig 4. X ray showing deficient abdominal wall more on right.

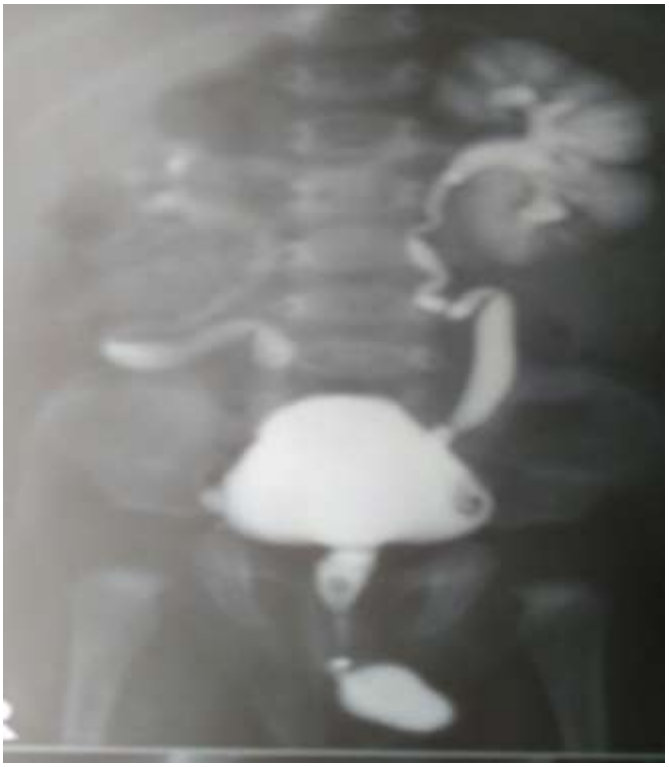

Fig 5. MCU showing bilateral VUR with PUV(posterior urethral valve).

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