

# Prune Belly syndrome: A rare case report

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## Abstract

Prune Belly syndrome (PBS) is a rare congenital anomaly of uncertain etiology almost exclusive to males. We report a case of term male baby born to a 39-year-old grand multipara with previous four normal vaginal births. There was no history of genetic or congenital anomaly in her family. Examination of the baby revealed hypotonia, deficient abdominal muscle, cryptorchidism, palpable kidney, and bladder. Ultrasound examination of the abdomen revealed bilateral gross hydronephrosis and megaureter. Provisional diagnosis of PBS was made and the baby was admitted in neonatal intensive care units for further management. Routine antenatal care with ultrasonography will help in detecting renal anomalies, which can be followed postnatally. Early diagnosis of this syndrome and determining its optimal treatment are very important in helping to avoid its fatal course.

**Key words:** Cryptorchidism, hydronephrosis, megaureter, Prune Belly syndrome

## INTRODUCTION

Prune Belly syndrome (PBS) is a rare congenital disorder affecting about 1 in 30,000 births<sup>[1]</sup> of which about 96% of those affected are male.<sup>[2]</sup> 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, megaureter and megacystitis.<sup>[2]</sup> The exact etiology of PBS is not known; however, some of the studies reveal the possibility of genetic inheritance and possible association with trisomy 18 and 21.<sup>[2,3]</sup> The prognosis of babies with PBS is poor with stillbirths and early infant deaths being common. We report a case of male baby with PBS because of its rarity and term birth.

## CASE REPORT

A 39 years old unbooked G<sub>5</sub>P<sub>4</sub>L<sub>4</sub>A<sub>0</sub> at 39 weeks 5 days of gestation was admitted to our labor room complaining of labor pain. She had four previous normal vaginal deliveries. There was no history of prior antenatal care and she belonged to a tribal community with lower socioeconomic status. There was history of consanguineous marriage with her husband who was 42 years old and a daily laborer. There was no history of drug intake, radiation exposure or fever with rashes in this pregnancy. Neither was she a known case of diabetes nor was there any history of genetic or congenital anomaly in her family.

On examination, she was in the second stage of labor

in cephalic presentation and regular fetal heart rate with fully dilated and taken up cervix. She delivered a term 2700 g female baby and the Apgar score was 7 and 9 at 1 and 5 min. Physical examination of the infant showed hypotonia. Abdominal examination revealed a distended abdomen with thin wrinkled skin and visible peristalsis [Figure 1] with palpable kidneys and bladder. Scrotal skin showed little rugae and no testes in the sac. Other systemic examination revealed no abnormality. Ultrasonography of the abdomen showed bilateral gross hydronephrosis with megaureter. The baby was referred to pediatric surgery department of higher center for further management.

## 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.<sup>[2]</sup> The etiology of PBS is unclear and possible familial genetic inheritance was reported in some of the studies.<sup>[3]</sup> Haeri *et al.* in 2010<sup>[4]</sup> 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. History of consanguinity and the mother being elderly gravida indicate the hereditary etiology in our case. The massive bladder distension and urinary ascites due to severe obstructive uropathy leads to

degeneration of the abdominal wall musculature and failure of testicular descent. The impaired elimination of urine from the bladder leads to oligohydramnios and pulmonary hypoplasia.<sup>[2]</sup>

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<sup>[5]</sup> 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<sup>[6]</sup> 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. PBS is rare in females, with fewer than 30 cases reported in the literature.<sup>[7]</sup> More complex morphogenesis of the male urethra may be the possible cause of obstructive anomalies at several levels indicating the higher incidence of this syndrome in males. After clinical examination, ultrasonography of abdomen, plain X-ray, intravenous pyelography and micturating cystourethrography can confirm the diagnosis.

Our patient comes under category 2 PBS as described by Woodard in 1985 [Table 1].<sup>[8]</sup> Management requires proper co-ordination among neonatology, nephrology, urology and other departments. Further management options include voiding cystourethrography with antibiotic cover in suspected cases of renal insufficiency or bladder outlet obstruction (BOO), suprapubic catheter in BOO to prevent urinary tract infections (UTI), orchidopexy and chest X-ray to rule out pneumothorax, pulmonary



**Figure 1:** Prune Belly syndrome: Distended abdomen, thin wrinkled skin with visible peristalsis

**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 No pulmonary hypoplasia

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hypoplasia and pneumomediastinum. In category 2 patients with renal insufficiency; reduction cystoplasty, ureteric shortening tapering and reimplantation with or without abdominoplasty (Monfort technique) may be performed. Careful surveillance with intervention for repeated febrile UTI or renal deterioration is also recommended.

The prognosis of PBS is usually poor as many infants are either stillborn or die within the first few weeks of life due to pulmonary hypoplasia or renal failure or a combination of congenital anomalies. Chronic failure is the most common complication and found in 25-30% of cases.<sup>[2]</sup> Diao *et al.* have reported renal failure to be the main cause of death in PBS.<sup>[9]</sup> Serum creatinine levels is a useful predictor of long term renal function and levels less than 60  $\mu\text{mol/l}$  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.<sup>[10]</sup> 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 Leeners *et al.*<sup>[11]</sup>

## CONCLUSION

PBS is a rare congenital anomaly which has no known prevention other than the routine use of screening for fetal anomalies. Routine antenatal care with ultrasonography will help in detecting renal anomalies early and optimal treatment thereafter can avoid fatal course of PBS.

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