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# Associated rare anomalies in prune belly syndrome: A case report

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ABSTRACT

The triad of deficient abdominal wall musculature, undescended testes and urinary tract anomalies characterizes the Prune Belly Syndrome (PBS). PBS can be associated with other comorbid urological and non urological conditions. But the full pathogenesis and best treatment is still a matter of debate. A term newborn with a classical PBS (Woodhouse Group 2, Smith and Woodard Group 2) plus lung hypoplasia and funnel chest deformity, a megapenis with a tight phimosis and an obturated anterior urethra is presented. Unfortunately, the baby died in urosepsis and renal failure in his 3rd week of life, despite urine drainage surgery and peritoneal dialysis undertaken. According to the best of our knowledge, this is an unique combination of rare anomalies in PBS patients.



Congenital absence of the abdominal wall musculature was described in 1839 by Fröhlich for the first time. Later in 1895, Parker made the first association with urinary tract anomalies, and Osler was credited to name it “prune-belly.” In 1950, Eagle and Barrett defined this syndrome as the triad of deficient abdominal wall musculature, undescended testes and urinary tract anomalies [1–6]. For the next half century, the syndrome was mainly regarded as a medical oddity, and most of the patients died due to overwhelming infections and destruction of their urinary tracts [3].

In its original form, PBS comprises complex malformations of the urinary tract, bilateral cryptorchism, and absence of the anterior abdominal wall muscles [2,5–11]. Sometimes, it is called the “Eagle-Barrett Syndrome” [7]. Examples seen with abdominal wall muscles present to a varying degree, and incomplete or even absent cryptorchism are called “pseudoprunes,” indeed. However, this term should not be mistaken in a way, that “good muscles necessarily mean good urinary tract” [8,10].

The estimated incidence of PBS in live births is 1 in 29 000 to 50 000, respectively 3.8 per 100 000 male live births. PBS occurs almost exclusively in boys, reports in females are only found anecdotal in medical literature reviews [2,5–7,11–14]. Affected girls usually do not exhibit the characteristic urogenital dysplasia, nor, of course cryptorchism [14].

The pathogenesis of PBS is still an unsettled controversy with the following predominating theories:

The first theory proposes a prenatal obstruction of the urinary tract, which causes urinary tract dilatation, fetal abdominal distension, and subsequent muscle wall hypoplasia and cryptorchism [2–6,13,14].

The second, embryology-based theory, proposes the failure of primary mesodermal differentiation between the 6th and 10th week of gestational age (GA), which leads to the defective muscularization of both, the abdominal wall and the urinary tract [2–6,13,14].

A third one, the yolk sac theory, indeed, proposes a dysgenesis of the yolk sac and allantois as causative for PBS [2].

Although each theory explains some elements of the syndrome, each one fails to explain others. In the first theory, the early congenital obstruction of the fetal bladder is thought to produce an extrapelvic mass, which compresses the developing abdominal wall. Such an explanation fits with the frequent finding of lower and central abdominal muscle loss, which spares out more lateral and upper abdominal parts. Yet, this theory fails to explain why male infants born with posterior urethral valves and most likely similar bladder enlargement are never found to have any congenital deficiency of abdominal muscles as well [3,6]. Urethral obstruction theory can also not explain the other complex morphological abnormalities of PBS [5], like absence of postobstructive changes (i.e. smooth muscle thickening, bladder wall trabeculation, and hypertrophy), or the severity of renal dysgenesis and dysplasia, exceeding the regular degree seen in hydroureteronephrosis and obstruction. And, any mechanical obstruction is an untenable explanation for a testicular maldescent [10].

Regarding embryology theory, the abundance of fibrous tissue with sparsely placed smooth muscle throughout the entire urinary

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tract is more indicative of an inherent problem with mesoderm differentiation than with obstruction. In addition, this theory provides a better explanation for the frequent association of this syndrome with a megalourethra [2,6]. It seems to explain the complexity of other morphological changes better as well, but it still remains insufficient to elucidate any causes and mechanisms of the underlying abnormalities of the genitals and upper urinary tract [5].

The third theory indeed, might explain the changes seen in the bladder and prostatic urethra, but it does not provide any explanations for the abnormalities seen in the upper urinary tract or testes [2,6].

Last but not least, Bellah and co-workers mentioned, that the existence of a "pseudoprune belly syndrome" (PBS uropathy, normal abdominal wall examination, and incomplete or absent cryptorchism) weakens the "in utero obstructive-theory" in favor of the "primary embryologic disturbance-theory" in the development of PBS [10]. Adebonoja added, that there is support to believe, that the PBS in its various forms, represents the manifestation of an "embryopathic insult" in between the 5th to 10th week of gestation. According to this theory it might be possible to explain the multiple system involvements, however, the exact nature of the insult is still open to speculation [1].

Most of the cases occur sporadically in children with a normal karyotype [5,8]. But, since a single explanation is not sufficient enough to describe the entire spectrum of PBS features alone, some other mechanisms and etiologies might be causative [6,15]. Like a single-gene abnormality or chromosomal defect. This is supported by the high incidence of PBSs found in association with either trisomy 13, trisomy 18 [5,6] or trisomy 21 [5,6,16,17]. After reviewing familial cases of PBS, other authors suggested a sex-influenced autosomal recessive mode of inheritance [15]. Reports of PBS associated with Turner syndrome (45 XO) support such a proposal as well [5,6,8]. Chromosome 17q12 deletions can be determinate in PBS patients [11,13] among others [5], too. Shah et al. reported a term, small for gestational age neonate expressing the full spectrum of a VACTERL (*v*ertebra, *a*nus, *c*ardiac, *t*rachea-*e*sophageal fistula, *r*enal, *l*imb) association together with a PBS triad. Such a combination is extremely rare and according to the authors in general incompatible with life. The concurrence of these two syndromes could lie in the common etiology of such defects in both, cranial and caudal mesodermal differentiation [18].

In general, PBS can be associated with pulmonary, cardiac, digestive, osteoarticular, or even other malformations [1,2,5–7,9,11,12,14,18,19]. Such non urologic comorbid conditions are reported by Routh et al. for cardiovascular in 25%, gastrointestinal in 24%, musculoskeletal in 23%, and respiratory in 58% of their cases. Comorbid renal and urologic conditions are reported in 53%, respectively. Sepsis, especially urosepsis are found in 14% and 2% [9]. Penis anomalies are found in 1% [9], namely penile urethral anomalies or phimosis are reported by others in anectodal case reports, too [2,4,11,15]. Lung hypoplasia, either alone [5–7,12], or in combination with pectus excavatum deformity [6,10,11,14,19,20] are found in literature as well.

The diagnosis of a PBS can be made earliest by prenatal ultrasound at 12 weeks of GA. Usually, a lower abdominal cystic echo caused by the abnormal dilatation of the bladder, reduced amniotic fluid, plus bilateral hydronephrosis, and absent abdominal muscles are present [20,21]. Postnatal, the following sonomorphologic classification is applied: grade I° – dysplastic kidneys with no appreciable surrounding renal parenchyma; grade II° – marked dilatation of the ureter and mild or no dilatation of the renal pelvis and calyces; grade III° – milder involvement ranging from the sonographic findings typical of grade II to those of a normal appearing urinary tract [22]. Urinary tract ultrasound, before and after voiding, is recognized as the best measure of dilatation [8].

In PBS uropathy voiding cysturethrography typically shows a funneling, unobstructed dilated posterior urethra, smooth-walled megacystis, and a high grade vesicourethral reflux. IV urography reveals dysmorphic (non-functioning or poorly functioning) kidneys with a severe hydroureteronephrosis [10]. But it has to be kept in mind, that any IV urography in the neonate has disappointing results, although it does give valuable anatomical information in the larger child [8]. Nevertheless, the radiological appearance of the urinary tract in PBS iv urography is always variable, and it is essential to realize, that this very dilated and bizarre appearance is not necessarily a sign of obstruction. And, if real obstruction is present, this is most often found in the membraneous part of the urethra. The prostatic urethra, indeed, is nearly always dilated, at least partly, because of the incomplete prostatic development seen in every PBS. The remainder of the urethra shows a variable degree of narrowing [9]. In severely affected patients it may be completely atretic. In milder cases the urethra often appears to be of normal caliber, but yet still inadequate for complete bladder emptying. The term "functional obstruction" seems to be the appropriate one, if the urethra produces more resistance than the weak detrusor can overcome [8].

The early assessment of the renal function is made by the measurement of plasma urea and creatinine concentrations, chromium EDTA clearance, and a DMSA scan [8].

In the neonatal period PBS is classified into three groups in accordance to the status of the urinary tract: In Group 1, the most severely affected (often with hypoplastic or atretic urethra) early death is inevitable. In Group 2, the children are ill as neonates (infection and gross dilatation of urinary tract), high diversion is often required and later reconstruction may be possible. Group 3 patients are healthy as neonates and little reconstructive surgery is required (good renal function despite very abnormal radiology). This classification forms the basis of many current management strategies [8].

Regarding its clinical manifestations the syndrome presents with: i) the non-viable oliguric form with severe kidney dysplasia, ii) a serious form consisting of marked renal dysplasia with megaureter, mega-vesicle and progressive renal failure, and iii) the more favorable form with moderate renal dysplasia and different degrees of enlargement of the ureters and bladder [14].

In the Smith and Woodard classification, indeed, the categories roughly correlate with the degree of renal dysplasia present: Group I patients usually have both severe renal dysplasia and pulmonary hypoplasia and will not survive. No urologic intervention will rescue these patients. Group II have intermediate degrees of dysplasia and renal function. These patients typically have full-blown features of the syndrome and a stagnant urinary system. Clearly, some of these patients will progress to renal insufficiency with our without any surgical intervention. In Group III, patients generally have a mild or incomplete form of the syndrome and well-preserved renal function. Although some of these patients may have impressive dilatation of the urinary tract relative to the patients without the syndrome, early reconstruction may be deferred, until there are free of infection [6].

High urinary diversion is still key in the management of any rapid renal deterioration in PBS patients to allow renal function to stabilize. If this fails to improve function, the infant has not enough sufficient renal parenchyma to support his/her life. Originally, suprapubic cystostomy and nephrostomy are extensively used. Subsequently, urinary diversion became more popular, but only to give way to a total surgical reconstruction [3,6,8,9,14]. Recently, urodynamic studies suggested that conservative nonsurgical management can be satisfactory and a number of authors have adopted this course in their individual management plans [3,6,8,9,14].