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A Case of Prune Belly Syndrome: Radiological Aspects of a Rare Disease

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Abstract **Case Report**

Prune Belly Syndrome (PBS) is a rare but morbid entity usually found in male neonates. It is classically defined by a triad of cardinal features that includes deficient abdominal wall musculature, urinary tract malformation and in males, cryptorchidism. Most of the patients have associated pulmonary, cardiac, skeletal and gastrointestinal tract anomalies. Children often require numerous surgical interventions including bilateral orchidopexy as well as individually tailored urinary tract and abdominal wall reconstruction. The prognosis of infants with PBS may be improved by quality antenatal follow-up, to enable the early diagnosis and preparation for prompt surgical intervention. We describe a case of PBS and emphasize the role of imaging, in the process of diagnosis and briefly review the subject.

Keywords: Prune Belly Syndrome (PBS), male neonates, pulmonary, abdominal distension.

INTRODUCTION

PBS is a rare congenital disorder that was first described by Frolish in 1839 [1]. It is also called Eagle-Barrett syndrome, Orbinsky syndrome, abdominal musculation syndrome and triad syndrome [2]. Male infants are most affected in close to 95% of cases and females represent less than 5% [3, 4]. The diagnosis is based on a radio-clinical triad that comprises deficient or absent abdominal wall musculature, bilateral intra-abdominal cryptorchidism and varying degrees of urinary system involvement such as ureterohydronephrosis, renal dysplasia, and megabladder [5, 6]. Patients with partial or unilateral abdominal wall deficiency, unilateral undescended testis, and female neonates with abdominal wall laxity are classified as Pseudo Prune Belly syndrome [7, 8]. A wide variability in disease severity exists with some patients also experiencing other associated defects including pulmonary hypoplasia, cardiac defects, imperforate anus, and intestinal malrotation [9, 10]. The urinary tract anomaly appears to be the most important determinant of survival in these patients [2]. No best practice guidelines exist for management of PBS owing to the scarcity and complex nature of the disease, along with diverse multisystem co-morbidities [11]. Therefore, multidisciplinary management is necessary and should be tapered to the needs of each individual child [12]. So far, reports on the role of imaging modalities remain relatively sparse. We describe in our work a case of PBS and emphasize the role of imaging, not only in prompting the diagnosis, but also in confirming it.

CASE REPORT

This is the case of a 6-year-old male infant who has been born by normal vaginal delivery to a non-consanguineous couple. His parents reported many episodes of dysuria, daytime and nocturnal urine leakage. On clinical examination, inspection of the unclothed infant showed abdominal distension, thin and slightly wrinkled appearance of the abdominal wall (Figure 1). Palpation reveals laxity and severe atrophy of the anterolateral abdominal wall allowing subcutaneous palpation of the intra-abdominal organs. Examination of the bursae revealed that they were empty. The rest of the physical examination was unremarkable. The paraclinical exams done comprised an abdominopelvic ultrasound (Figure 2) which showed features in favor of major bilateral ureterohydronephrosis associated with a thickened-walled diverticular megabladder, and impalpable bilateral testes within the scrota. Voiding cysto-urethrography (Figure 3) demonstrated bladder diverticulum, bilateral grade V vesicoureteral reflux with tortuous and dilated ureters. When communicating results with the child's parents, they announced that he had previously benefited from an abdominal computed tomography (Figure 4) that revealed major bilateral ureterohydronephrosis associated with a megabladder, aplasia of the muscles of the anterior abdominal wall. The entire spectrum of radiological and imaging features

was consistent with PBS. No clinical or radiological evidence for any known associated congenital anomalies was found.

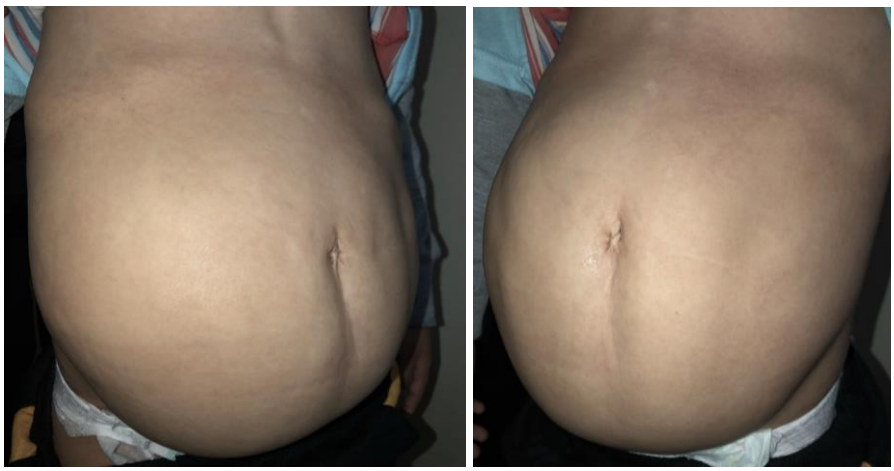

Figure 1: Clinical picture of the infant showing abdominal distension with thin and slightly wrinkled appearance of the abdominal wall

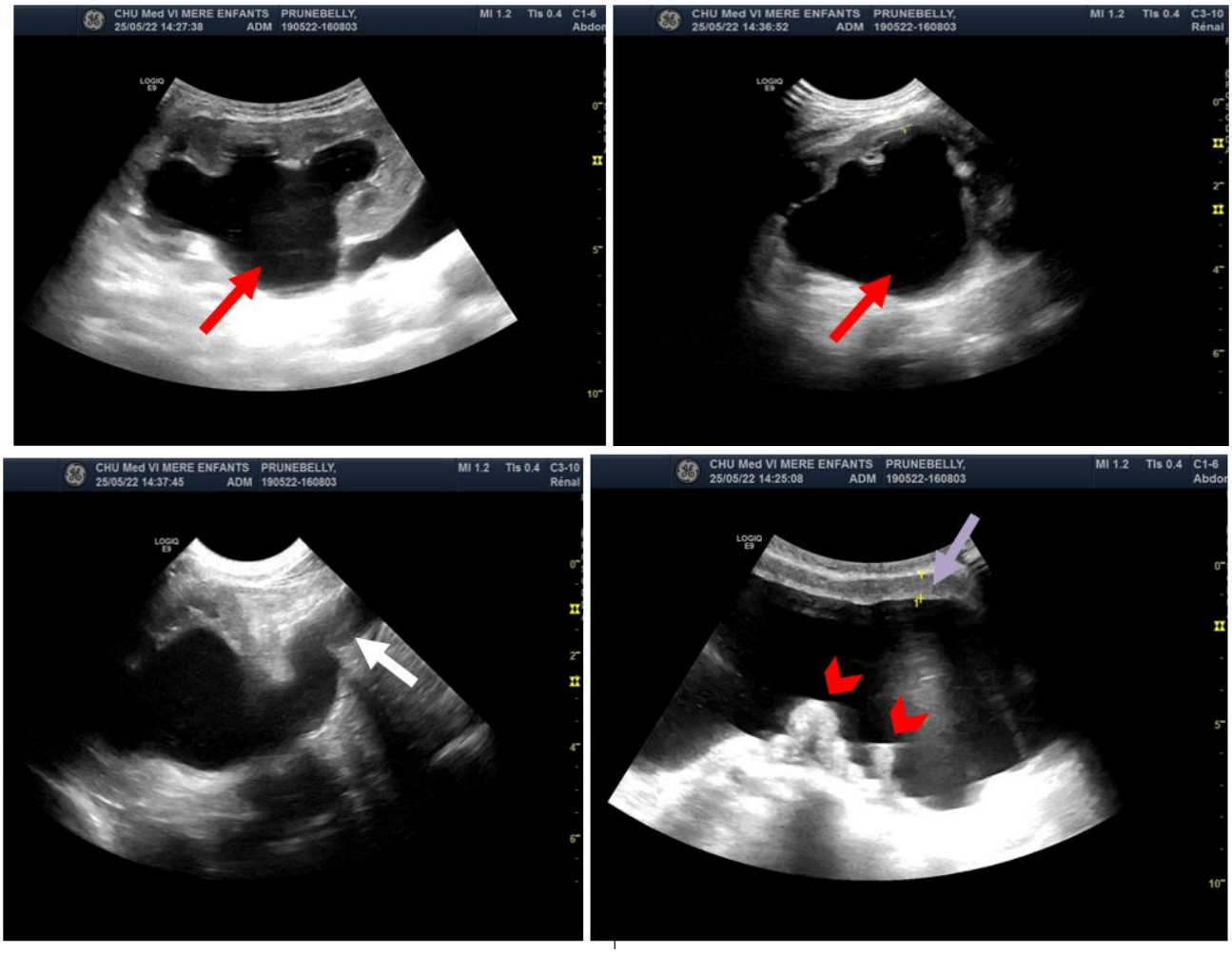

Figure 2: Abdominopelvic ultrasound showing major bilateral ureterohydronephrosis (red arrows), with tortuous appearance of ureters (white arrow), associated with a thickened-walled (yellow arrow) diverticular megabladder (red head arrows)

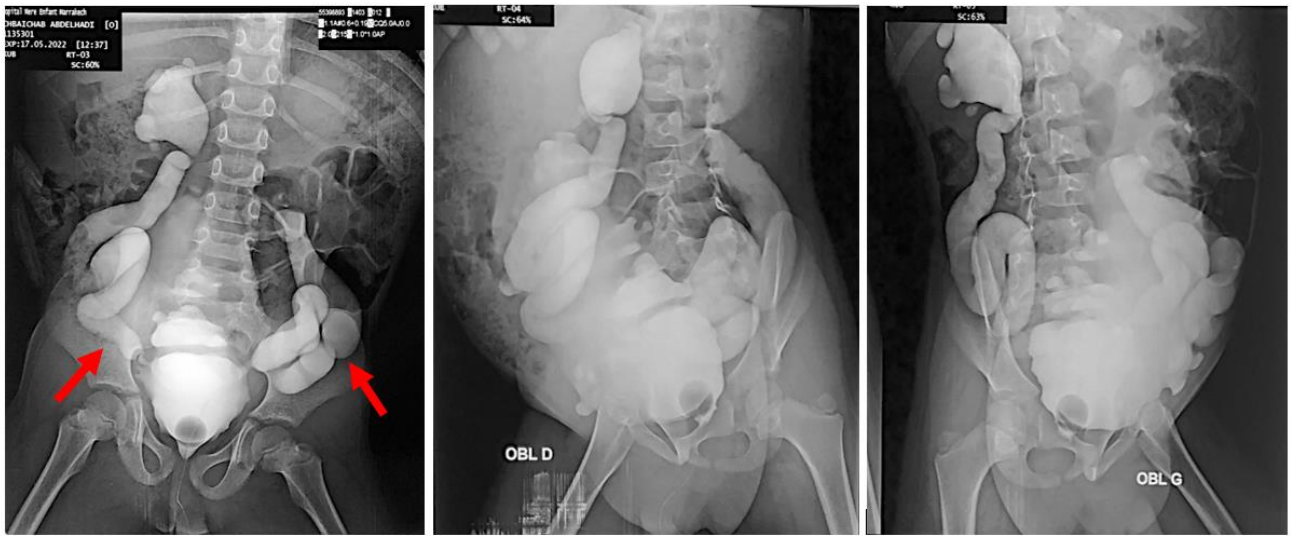

Figure 3: Voiding cysto-urethrography demonstrating bladder diverticulum, bilateral grade V vesicoureteral reflux, with tortuous and dilated ureters (red arrows)

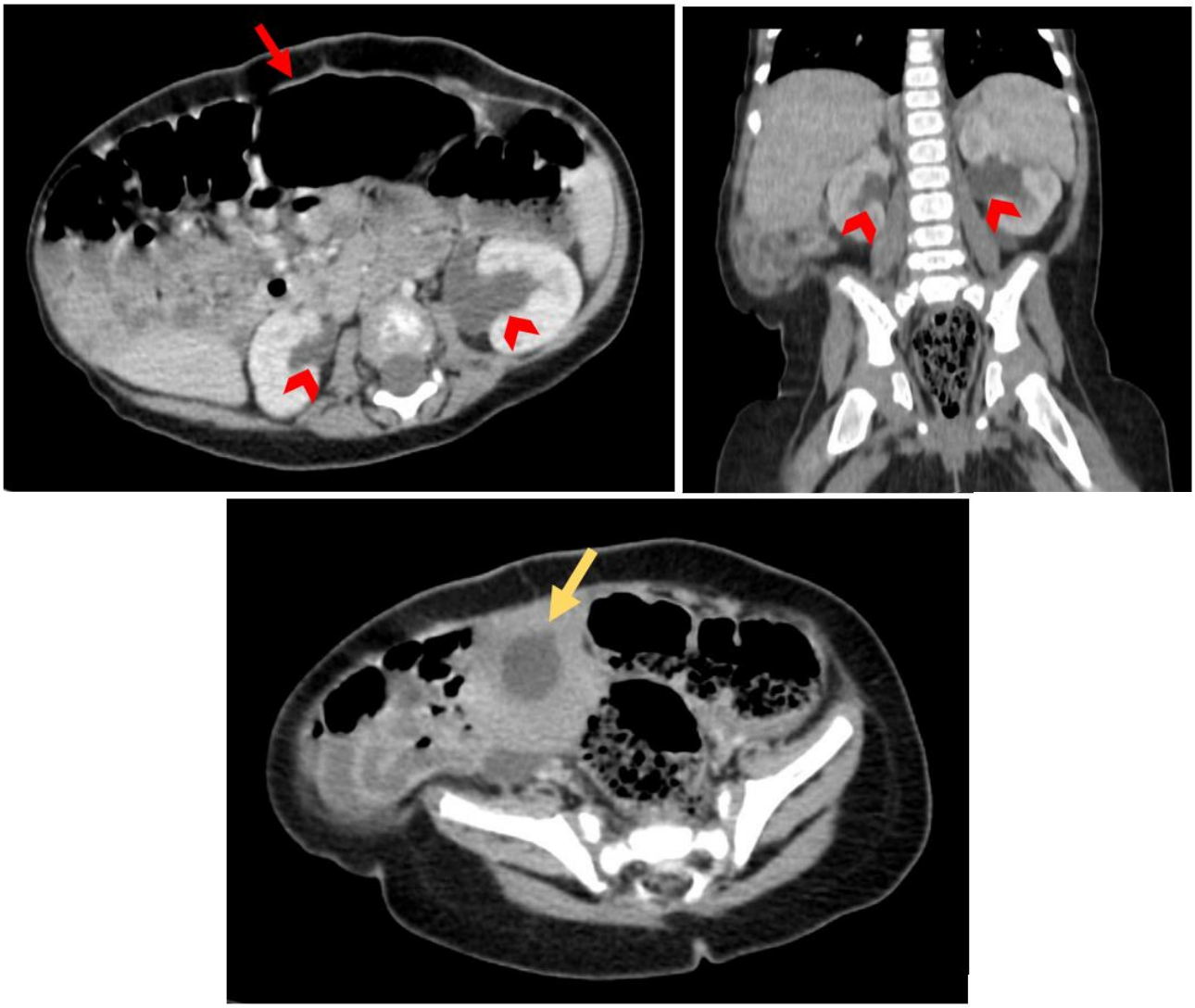

Figure 4: Abdominal CT-scan revealing aplasia of the muscles of the anterior abdominal wall (red arrow), major bilateral ureterohydronephrosis (red head arrows), associated with thickened bladder wall (yellow arrow)

DISCUSSION

PBS is a rare, congenital and complex disease affecting about 1 in 30.000 to 40.000 live births [13, 14]. The term "prune-belly" reflects the characteristic wrinkled, crumpled and loose appearance of the abdominal wall due to the complete or partial absence of abdominal wall muscles, reminiscent of a "prune" [7, 15]. PBS is classically defined by a triad of cardinal features including deficient or absent abdominal wall musculatures, bilateral cryptorchidism and urinary tract abnormalities [4,16]. The real etiology is largely unknown. Several theories have been formulated, including the hypothesis of prenatal urine obstruction, the embryological hypothesis over a primary mesodermic differentiation between the 6th and the 10th weeks of gestation which could be responsible for a defective urinary tract and abdominal musculature. A third hypothesis is the yolk sac theory, which proposes a dysgenesis of the yolk sac and allantois as the basis of the PBS [10, 17]. The PBS predominates in male infants as it was the case with our patient. While the partial or pseudo-PBS is more common in female infants, who rarely manifest urinary symptoms. Based on the male sex predominance, a genetic influence has eventually been suggested with a possible transmission through autosomal recessive sex-linked genes [18, 19]. A classification system which scored PBS children into one of three categories of phenotypic severity in the neonatal period was proposed by Woodard which helps prioritize clinical management and care. Category I represents neonates who experienced marked oligohydramnios secondary to severe renal dysplasia or functional bladder obstruction, with resultant pulmonary hypoplasia and Potter's features. Most children are stillborn, and newborns remain alive for a few days until death. Patients with Category II present the full spectrum of the syndrome with the classic triad. Category III patients present mild or incomplete forms of PBS, with less severe renal alterations as well as normal pulmonary function [14, 20]. More recently, Wong et al. expanded on Woodard's original classification by designing a severity scoring system referred to as RUBACE, an acronym based on six subscores—renal, ureter, bladder/outlet, abdominal wall, cryptorchidism, and extragenitourinary [21].

On physical examination, the anterior abdominal wall is usually thin with visible bowel peristaltic activities. All of the muscle layers below the level of the umbilicus usually are affected, where fibrous tissue rather than muscle tissue may be present [13, 21]. Bilateral cryptorchidism is one of the three cardinal signs defining the syndrome, and is most often bilateral [1]. The main clinical signs of the PBS are urinary tract malformations including renal dysplasia, megabladder, dilatation, stenosis or atresia of the ureters, polycystic kidneys, hydronephrosis, and sometimes vesicoureteral or urethral diverticulum [18]. Vesicoureteral reflux is present in approximately two-thirds of PBS patients, with often dilated, elongated, and tortuous ureters [21]. Therefore, the assessment of the kidney function seems fundamental for the prognosis. Many other malformations may be associated with PBS. In fact, it is a multisystem disease, with patients displaying cardiopulmonary, gastrointestinal, and musculoskeletal anomalies to a varying degree [17]. Prenatal diagnosis plays a key role in early detection of PBS based on obstetric ultrasound, since it would allow prompt management of newborns with the syndrome at birth, thus resulting in improved survival [22]. Indeed, it can usually be diagnosed as early as 12 weeks of gestation. An antenatal diagnosis should be considered whenever the following ultrasound anomalies are evident: oligohydramnios, significant bladder dilatation, pyelocaliceal dilatation and abdominal wall weakness with prominence on sagittal sections [1, 9, 17]. In the postnatal period, the diagnosis is made by an abdominopelvic ultrasound. In typical PBS, the kidneys show various degrees of dysplasia, cystic dilatation of the calices, and dilated, enlarged, and tortuous ureters. In addition to dilatation of the renal pelvis and the ureters, bladder filling and bladder wall thickness can be determined. The first additional radiological examination performed in patients with prune belly syndrome should be voiding cystourethrography, an investigation that is independent of the degree of renal function and gives valuable information regarding the lower urinary tract (bladder outflow) and whether vesicoureteral reflux is present [19].

The search for possible associated malformations may require, depending on the case, abdominopelvic computed tomography (CT) scan, trans-thoracic echography, plain X-ray, karyotype and renal assessments. The Karyotype helps to investigate deletion or suppression on the Hepatocyte Nuclear Factor 1-beta (HNF-1β) which may be responsible for multisystem defects in humans [1, 8]. The management of PBS is quite complex [1]. Caring for patients with PBS requires a large multidisciplinary team approach to help these children thrive, gain weight, and be prepared for eventual surgery [14]. Opinions are divided between surgical and conservative treatment [1, 23]. However, the mainstays of treatment are abdominoplasty, a cure for cryptorchidism with lowering of the testicles and orchidopexy, and urinary tract reconstruction [1, 8]. Abdominoplasty is indicated to improve aesthetics, and intra-abdominal pressure improves bladder emptying [20, 21]. For patients who are affected with mild abdominal muscles hypoplasia, postures may be acceptable. However, for severe cases, surgical intervention must be envisaged [8, 24]. Correction of cryptorchidism and phimosis should ideally occur between 6 to 18 months due to recurrent urinary tract infections and deterioration of renal function. In the other hand, delayed orchidopexy is known to contribute to infertility [20]. Kidney transplantation is often unavoidable for patients with renal insufficiency or advanced failure [21]. As for conservative management, prophylactic antibiotics should be administered to reduce

the risk of urinary tract infection. Hemodialysis and peritoneal dialysis are options for renal replacement therapy in patients with chronic kidney disease [21]. Not surprisingly, PBS has been shown to profoundly affect quality of life in pediatric patients, negatively impacting their physical, emotional, social and school functioning [11]. The prognosis for patients with PBS varies according to the severity of pulmonary hypoplasia and urinary tract abnormalities. Pulmonary hypoplasia is the main reason patients cannot survive the neonatal period. The severity of the urinary tract abnormalities and renal function determine not only the mortalities but also the long-term outcome. Therefore, a comprehensive approach that includes rehabilitation, early stimulation and psychological support should be considered in these patients [9, 16, 19, 24].

CONCLUSION

PBS is a rare entity mainly affects males, with a broad spectrum of symptoms involving the abdominal wall, the testicles and the urinary tract. The role of the radiologist remains crucial in the diagnostic work-up and in follow-up. Early diagnosis with ultrasound allows not only for prompt multidisciplinary management, but also allows termination of an undesired pregnancy. Thus, increased awareness of all the aspects of this disease is essential, especially in large maternal and pediatric centers.

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