Prune belly syndrome with urethral hypoplasia and vesico-cutaneous fistula: A case report and review of literature

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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.\(^1\) Although the etiology of PBS has not yet been well defined, some hypotheses have been suggested such as distal obstructive uropathy and mesodermal defect of the abdominal wall and the urinary tract.\(^1\)\(^3\)

Urethral hypoplasia or atresia 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 vesico-cutaneous fistula. Herein, we report a case of PBS with urethral hypoplasia associated with vesico-cutaneous fistula.

CASE REPORT

A 35 weeks gestational age male neonate was referred to pediatric urology 6 hours after delivery with a history of prenatal bilateral hydroureteronephrosis, megacystis, oligohydramnios and inability to void postnatally. Clinical examination revealed typical appearance of PBS with no dysmorphic facies. Abdominal examination showed a small cutaneous fistula opening below the umbilicus pouring urine and distended urinary bladder. Genital examination showed a small phallus with bilateral non-palpable testes and empty scrotum [Figure 1].

Trial of insertion of urethral catheter was unsuccessful. Laboratory investigation revealed Hgb of 13 gm/dl and Creatinine of 43 µmol/l. Ultrasound revealed marked grade IV bilateral hydronephrosis with dilated tortuous ureters down to a semi-filled urinary bladder.

On the 2\(^{nd}\) day of life, the patient underwent examination under anesthesia. Trial cysto-urethroscopy using an infant 7.5 F scope was unsuccessful. So, a cutaneous vesicostomy (Blocksom...
technique) was performed after excision of the vesico-cutaneous fistula [Figure 2].

Voiding Cystourethrogram (VCUG) postoperatively showed bilateral high grade vesico-ureteric reflux (VUR) with dilated posterior urethra and atretic anterior urethra [Figure 3].

The child general condition was stable and he was discharged on prophylactic antibiotics for his high grade VUR with acceptable creatinine level of 36 µmol/l.

**DISCUSSION**

Prune belly syndrome has 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. However, survivors will have a varying degree of renal affection throughout their lives. Our patient had the chance of urinary drainage via a spontaneous vesico-cutaneous fistula which maintained partially his amniotic fluid.

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. Unless it is associated with a patent urachus or a spontaneous vesicocutaneous fistula, urethral atresia is often lethal. 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 and this was probably the scenario of what happened in our patient.

There are only few reports about the association between PBS and urethral hypoplasia in the literature. Reinberg and colleagues 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 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.

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). In our patient we did the same procedure of diversion aiming at decompression of the upper tract and improvement of the future renal function.

Treatment of urethral hypoplasia in PBS includes gradual progressive dilation using soft catheter with insertion of stent, urethrotomy and open reconstruction. However, conflicting data have been reported [Table 1].

Passerini-Glazel and colleagues (1988) 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), however, this technique is not uniformly successful and may require a more formal urethroplasty with skin flaps or grafts, or both.

Recently, Kajbafzadeh et al., 2010 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.

In our patient, the management of urethral hypoplasia will be dictated according to the outcome of renal function and future condition of his upper and lower urinary tract.

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Table 1: Literature summary of patients with PBS and urethral hypoplasia

| Reference                  | Number of cases | Method of repair | Fate                  |
|----------------------------|-----------------|------------------|-----------------------|
| Passerini-Glazel et al.,   | 5               | Progressive urethral dilatation | Successful in all |
| Reinberg et al.,[4]        | 5 (2 survived) | Perineal Urothrotomy+ open reconstruction in 1 | Successful |
| González et al.,[7]        | 5               | Progressive urethral dilatation | Successful in 2 |
| Kajbafzadeh et al.,[8]     | 7               | Urethral hydro distension | Successful in all |

PBS: Prune belly syndrome

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