Prune belly syndrome (PBS) is a rare congenital disorder characterized by deficient abdominal wall muscles, urinary tract malformation, and, in males, cryptorchidism. We present a case of PBS in China. The patient was a newborn baby boy who had wrinkled, “prune-like” abdominal skin, bilateral cryptorchidism, and urinary system malformation, complicated with hypoplasia of the lung and branch of the coronary artery—right ventricular fistula. His kidney function was inadequate. The patient subsequently died at age 28 days due to septicemia from a severe urinary tract infection.

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1. Introduction

Prune belly syndrome (PBS) is a congenital disorder that presents with three main features: abdominal wall muscular layer defect, urinary system malformation, and bilateral cryptorchidism. The incidence is 1:35,000 to 1:50,000 live births. Over 95% of patients are boys, and only 3–5% are girls. In girls, PBS is typically more severe because of a higher incidence of urethral atresia. Up to 75% of patients with PBS have associated pulmonary, skeletal, cardiac, and gastrointestinal defects. Pulmonary hypoplasia due to oligohydramnios secondary to renal dysplasia is a potential cause of death after birth, but the extent of urinary tract disease is the major prognostic factor.

2. Case Report

A 2-day-old boy was admitted for right bulging flank in November 2011. Gestational age was 39 weeks, with no fetal distress or asphyxia. The birth was by Cesarean section. Apgar scores were 8 at 1 minute and 9 at 5 minutes. Birth weight was 2900 g, birth length 50 cm, head
circumference 34 cm, and chest circumference 33 cm. The infant was born to a 20-year-old uncomplicated mother, Gravida 1 Para 1. Prenatal sonography showed an intra-abdominal cystic collection in the fetus at 28 weeks’ and 34 weeks’ gestation. There was no prenatal intervention. After admission, physical examination showed bulging flanks, especially on the right side, thin flabby, wrinkled skin (Figure 1), and bilateral testes impalpable within the scrotum. There were no other issues, and urine output was normal.

The chest and abdominal X-ray showed normal lung volume, hypoplasia of the upper mediastinum and lung, and no lung marking in the upper right lung, upper or middle left lung. Diffusely distended flanks were seen, with mass-like areas in the lower abdomen. Voiding cystourethrogram showed bladder diverticulum and bilateral vesicoureteral reflux, and the bilateral ureters were tortuous and dilated (Figure 2). An abdominal computed tomography (CT) scan showed small and irregular bilateral renal shape, renal parenchymal density, mild discordance of part of the renal pelvis, and dilatation of the renal calices and superior segments of both ureters. Cystic outward protrusion on the right side of the dilated bladder was also noted (Figure 3). Echocardiography showed mesocardia, branching of the coronary artery—right ventricular fistula, and mild tricuspid reflux. The urinalysis showed urine occult blood (+), protein (+), and white blood cell count (WBC) 32.8/μL (0–25); renal function showed blood urea nitrogen 12.94 mmol/L and creatinine 284.4 μmol/L on day 5. The baby had fever from day 7. Urine analysis showed urine occult blood (+), protein (+), and WBC 1997/μL (0–25); renal function showed blood urea nitrogen 21.83 mmol/L and creatinine 349.1 μmol/L on day 7. Urine culture was negative. Chromosome analysis was normal. The clinical diagnoses were PBS, coronary artery branch fistula, urinary tract infection, and renal inadequacy. The parents chose to cease treatment, and the patient died subsequently at 28 days due to septicemia from a severe urinary tract infection.

3. Discussion

PBS is also known as Eagle-Barrett syndrome, the triad syndrome, and urethral obstruction malformation sequence. The wrinkled, “prune-like” abdominal skin due to an abdominal wall muscle defect is a characteristic manifestation and the first clue of PBS at the neonatal physical examination. Urinary tract anomalies and bilateral cryptorchidism are the other two characteristic changes.

The exact etiology of PBS is unknown. The obstructive theory supports that severe bladder outlet or distal urethral obstruction existed early in gestation, causing bladder distention, ureteral dilatation, hydronephrosis and atrophy of the abdominal wall muscles, as well as undescended testicles.4,5 The mesenchymal defect theory proposes that early injury of the mesoderm results in the associated anomalies.6 Although most cases were reported sporadically and a variety of inheritance patterns have been proposed, Ranjith reviewed 11 cases of familial PBS and proposed a sex-influenced autosomal recessive inheritance pattern,7 however, the exact mode of inheritance of PBS is still unclear.

The characteristic outward feature, the abnormal abdominal appearance of a wrinkled, loose, “prune-like” abdomen, is due to an abdominal muscle defect. The deficiency is usually asymmetric. Sometimes, the huge bladder, dilated ureters, polycystic kidney, or hydronephrosis can be seen or touched through the abdominal wall. Malformation of the urinary system includes different degrees of kidney dysplasia, hydronephrosis, dilated and tortuous ureters, enlarged bladder, sometimes diverticulum near the vesicoureteric junction, and urethral obstruction. The urethra in patients with PBS may be
stentotic, dilated, or atretic, but females have a higher incidence of urethral atresia.\textsuperscript{2,8} Vesicoureteral reflux is present in two-thirds of patients.\textsuperscript{9} The condition of renal function and the severity of the effects of the urinary system are the prognostic determinants. The patient we report here had kidney dysplasia and renal function insufficiency, dilated and tortuous ureters, bladder diverticulum, and vesicoureteral reflux, and subsequently died at 28 days. Almost all male patients have cryptorchidism, mostly bilateral. Abnormalities of the cavernous or prosthetic hypoplasia have also been reported. For female patients, genital abnormalities include vaginal atresia, bicornuate uterus, and urogenital sinus. Men with PBS have normal sexual function if there are no cavernous abnormalities. There have never been reported instances of fertility, for either men or women, in this syndrome.\textsuperscript{8} Respiratory manifestations in PBS include hypoplasia of the lungs and cystic adenomatoid malformation. Significant clinical pulmonary problems are noted in 55\% of patients.\textsuperscript{10} Pulmonary hypoplasia results from oligohydramnios and can result in different degrees of respiratory insufficiency. It is also the main reason that patients cannot survive in the neonatal period. Crompton studied the respiratory function of 11 patients aged 7–19 years with PBS: he reported that abnormalities of lung function appeared to be secondary to the musculoskeletal disorder associated with PBS rather than to parenchymal lung disease.\textsuperscript{11} Cardiac anomalies are present in up to 10\% of patients. Patent ductus arteriosus, atrial septal defect, ventricular septal defect, and tetralogy of Fallot are common issues. Our patient had branching of the coronary artery—right ventricular fistula. This has not been reported previously. Gastrointestinal malformations were seen in 30\% of patients with PBS.\textsuperscript{10} Intestinal malrotation and mesenteric anomalies are frequent, including atresia, stenosis, volvulus, imperforate anus, splenic torsion, Hirschsprung disease, gastrochisis, and persistence of common fetal cloaca. Musculoskeletal involvement has been reported in 45\% of patients. These can be congenital malformations, including club feet, limb deficiencies, teratologic hip dysplasia, and vertebral malformations. They can also include postnatal problems such as renal osteodystrophy, scoliosis, pectus excavatum, and pectus cranium.\textsuperscript{12} Motor developmental delays due to poor abdominal musculature assisting in movement and balance, recurrent respiratory tract infection secondary to incomplete cough mechanism, and chronic constipation resulting from absent abdominal musculature are common in such patients.

Prenatal ultrasound is able to detect abnormalities of the urinary tract associated with the typical appearance of the abdominal wall. The postnatal diagnosis of PBS can be easily established. More and more prenatal diagnoses have been reported recently. Hosbino reported a case of prenatal diagnosis of PBS at 12 weeks of pregnancy.\textsuperscript{13} The therapeutic regimen for PBS includes respiratory support of life-threatening pulmonary hypoplasia and surgeries including abdominoplasty, orchiopexy, and urinary tract reconstruction. For patients with mild abdominal wall dysplasia, postures are acceptable and labor is not restricted. There is no need for an abdominoplasty. However, for severe cases, whether and when the surgeries should be performed is determined on a case by case basis. Pyelostomy, ureterostomy, and cystostomy are also undertaken to shunt the urine temporarily in some unstable infants who cannot tolerate an operation. Dialysis or renal transplantation is often unavoidable for patients with renal insufficiency or failure. Fetal intervention in PBS is not warranted except in rare instances of dystocia secondary to fetal bladder distention.\textsuperscript{14} There are reports that for patients with severe lower urinary tract obstruction, prenatal vesicoamniotic shunting results in a good outcome with preservation of renal and pulmonary function.\textsuperscript{14} Regardless of whether surgeries are undertaken or not, patients with PBS need permanent multidisciplinary medical care and close follow-up.

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. Woodhouse and coworkers reviewed 47 cases with PBS and classified them into three groups according to the state of the urinary tract in the neonatal period: they proposed that if the patients were severely affected and had worsening renal function after birth, early death was inevitable, but if the patients were well and had good renal function, then although they had the urinary tract abnormalities, three-quarters of them grew up normally with satisfactory renal function and health.\textsuperscript{15}
References