Case Report

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ABSTRACT

We describe a 2-year-old boy seen in Kenya who presented with fever, dysuria, foul smelling urine, and diarrhea. His previous ultrasonography showed an overdistended bladder and hydronephrosis. Physical examination revealed a distended abdomen, loose abdominal wall muscles with thin, flabby, and wrinkled skin on the abdominal wall, hypoplastic scrotum, and bilateral cryptorchidism. A diagnosis of prune belly syndrome was promptly made. The urinary tract infection was duly treated with intravenous ceftriaxone and the child followed by pediatric urology.

Key words: *Prune belly, Urinary tract infection, Cryptorchidism*

Prune belly syndrome (PBS) is a rare congenital disease, characterized by a triad of loose abdominal musculature, urinary tract system malformation, and, in males, bilateral cryptorchidism [1]. The condition was first described by Frolich in 1939 and the term "prune belly syndrome" was coined by Osler in 1901 [2]. We describe a 2-year-old boy with PBS – the diagnosis was not made until the child was seen by one of us (HKAY). Prompt clinical diagnosis is possible with recognition of the cardinal features of abdominal wall muscular layer defect, urinary tract malformation, and bilateral cryptorchidism [1].

CASE REPORT

A 2-year-old boy with a distended abdomen and loose abdominal wall musculature was admitted to the hospital in Kenya with a three days history of low-grade fever, painful urination, foul smelling urine, decreased urine output, and diarrhea. The urine was malodorous and cloudy. He had loose to watery stools three times a day. The child was irritable and had a poor appetite.

The child was born to an African mother at home following a normal spontaneous vaginal delivery in an African village. There was no prenatal intervention. There was no apparent abnormality noted at birth. He was first admitted to the hospital one year ago because of irritability, inconsolable crying, decreased urine output, and turbid urine. During that admission, abdominal ultrasonography revealed severe hydronephrosis and an overdistended bladder. He was treated as acute urinary retention with catheterization and oxybutynin hydrochloride, and was discharged home 5 days later.

On admission, the temperature was 36.0℃, heart rate 147 beats per minute, respiratory rate 20 breaths per minute, and oxygen saturation 100% on room air. The child was conscious but irritable and inconsolable. Physical examination performed by one of us (HKAY) revealed a boy with 5 to 10% dehydration manifested as

decreased urine output, decreased skin turgor, sunken eyes, and dry oral mucosa. The child was pale. He had loose abdominal wall musculature with thin, flabby, and wrinkled skin on the abdominal wall, and abdominal distension with bulging of the flanks, especially on the right side (Figure 1).

Figure 1 - Characteristic appearance of the winkled prune-like abdomen and bulging of the flanks, especially on the right side

There was no abdominal tenderness or guarding. Bowel sounds were normal. The bladder was not palpable. The scrotum was hypoplastic and the testes were impalpable. The rest of the physical examination was unremarkable. Based on the history and physical findings, a diagnosis of PBS was made.

The complete blood picture showed white cell count of $10.1x10^3$/µL with predominance of lymphocytes ($3.9x10^3$/µL); hemoglobin of 8.6 g/dL with mean corpuscular volume (MCV) of 57.2 fl and mean corpuscular hemoglobin (MCH) of 18.9 pg; and platelet count of $771x10^3$/µL. The urinalysis revealed bacteria and numerous white blood cells. Urine culture was not performed due to limited resources. Renal function revealed creatinine level of 178 pm/L (normal range is 50-100 pm/L).

The child was rehydrated with intravenous fluids. Intravenous ceftriaxone was given for treatment of the urinary tract infection. The child improved over the next few days. The child was referred to a pediatric urologist because of the undescended testes. Orchidopexy was subsequently performed.

DISCUSSION

Prune belly syndrome (PBS), also known as Eagle-Barrett syndrome, abdominal muscle deficiency syndrome, triad syndrome, and urethral obstruction malformation sequence is a rare congenital disorder with an estimated incidence of 3.8 cases per 100,000 live-births in all ethnicities. PBS is characterized by three cardinal features: abdominal wall muscular layer defect, urinary tract malformation, and, in males, bilateral cryptorchidism [1,3-5]. Ninety five percent of cases occur in males while only 5% of cases occur in females [1,3]. Most cases occur sporadically, although familial cases have rarely been reported.

PBS may be diagnosed antenatally by ultrasonography which may detect abnormalities of the urinary tract such as bilateral hydroureters and hydronephrosis, a distended, thin-wall bladder, oligohydramnios and the typical appearance of the abdominal wall [6]. However, in resource-limited setting such as Kenya, many parents may not have access to antenatal care. As such, the diagnosis may not be made prenatally.

The characteristic winkled, prune-like skin due to an abdominal wall muscular defect and bilateral cryptorchidism when noted in the neonatal period often gives clue to the diagnosis. However, the loose abdominal wall musculature might not be obvious at birth as is illustrated in this case. Hence, the diagnosis might be missed in the neonatal period. In such cases, the diagnosis is often made later in childhood when the child presents with a urinary tract infection and physical findings of loose abdominal musculature and bilateral cryptorchidism. Familiarity of this condition will allow a prompt diagnosis to be made.

The wrinkled, prune-like abdominal skin is characteristic. The abdominal wall typically consists of poorly organized central abdominal musculature interspersed with dense collagen. In

general, all muscles in the anterior abdominal wall below the umbilicus are affected with the muscles replaced by fibrous tissue. Characteristically, the muscles in peripheral abdominal wall are not affected. The abdominal muscular defect is often asymmetric with bulging of the flanks more on one side. Early abdominal wall reconstruction is important as improved abdominal tone will lead to improved sensation of bladder distension and enhanced efficiency of the Valsalva manoeuvre in bladder emptying [5].

Almost all male patients with PBS have cryptorchidism. The cryptorchidism is usually bilateral. Impaired fertility is a well known complication of cryptorchidism [7]. It is important that orchidopexy should be performed no later than 15 months of age so as to preserve function of the testes. Orchidopexy should be performed between 6 and 12 months of age, provided that a surgeon with pediatric training and experience performs the procedure [7]. Affected females, on the other hand, may have vaginal atresia, bicornuate uterus, and urogenital sinus.

An ectatic or low-pressure, non-obstructed, dilated urinary system, evident from the renal pelvis to the urethra is one of the hallmarks of PBS. A study investigating the urological complications of PBS found mega-bladder in 81.8% and hydronephrosis in 56.3% of patients [1,3,8]. The urinary bladder, although enlarged, is not trabeculated. Other malformations of the urinary system includes various degrees of renal dysplasia, dilated and tortuous ureters, diverticulum near the vesicoureteric junction, patent urachus or urachal diverticulm, and urethral obstruction. The urethra may be dilated, stenotic, or atretic. Vesicoureteric reflux is found in approximately 75% of patients with PBS.

Respiratory complications such as pulmonary hypoplasia are common [1,3]. Pulmonary hypoplasia is due to oligohydramnios secondary to renal dysplasia. The oligohydramnios may result in Potter's facies. Pulmonary hypoplasia is an important cause of mortality in the neonatal period. Cystic adenomatoid malformation and rib cage narrowing may also occur. Recurrent respiratory infection is common and can be attributed to incomplete cough mechanism.

Forty five percent of affected patients have musculoskeletal defects, such as club feet, digital anomalies, vertebral malformations, teratologic hip dysplasia, and limb deficiencies (lower limb hypoplasia or aplasia) [1,3,9]. Postnatal problems such as scoliosis, pectus excavatum, and pectus carinatum occur with increased frequency in patients with PBS.

Thirty percent of patients have gastrointestinal malformations such as Hirschsprung disease and imperforate anus [1,3,10]. Other gastrointestinal malformations include intestinal atresia or stenosis, malrotation of the intestine, gastroschisis, and persistence of common fetal cloaca. Twenty four percent of patients have coexisting congenital cardiovascular anomalies such as atrial septal defect, ventricular septal defect, patent ductus arteriosus, and tetralogy of Fallot [1].

Approximately one third of patients with PBS outside the postnatal period will progress to renal failure requiring transplantation [5,11]. It is important to detect PBS early so that urinary tract infections can be properly treated which help to minimize the renal complications and to preserve renal function [12]. The severity of the renal dysplasia is the main prognostic factor [8,11]. Woodhouse et al classified patients with PBS into 3 risk groups according to the state of the urinary tract in the neonatal period [13]. In the first group, patients were severely affected at birth with oliguria and worsening renal function. Early death was inevitable in this group of children. In the second group, patients became ill during the neonatal period with poor kidney function and grossly dilated urinary collecting system. In this group, death might be preventable if early and proper measures are instituted. In the third group, children remained healthy in the neonatal period with satisfactory renal function [10]. Prognosis is best for children in the third group. Our case belongs to the third group as the

child was otherwise healthy and had satisfactory renal function. In developing countries where medical facilities were not readily available, children in group 1 and group 2 were not expected to live beyond early infancy. Most patients with PBS in these countries who present with recurrent urinary tract infections in childhood belong to the third group. This has important implications on counseling parents that little, if any, reconstructive surgery may be necessary, and the key to management is to establish free drainage of the urinary tract and to prevent urinary tract infection [5,13].

This report highlights an important message in the management of PBS in a resource-limited setting. Prompt clinical diagnosis is possible with recognition of the cardinal features of PBS, namely, abdominal wall muscular defect, urinary tract malformation, and bilateral cryptorchidism.

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