Original Article

# Prune belly syndrome: A report of 15 cases from Sudan

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## ABSTRACT

Prune belly syndrome is a rare congenital malformation of unknown aetiology, composed of a triad of deficient abdominal wall muscle, cryptorchidism and urinary tract anomalies. The majority of patients have associated pulmonary, skeletal, cardiac, and gastrointestinal defects. This was a prospective, case finding study that was conducted in the main paediatric hospitals in Khartoum state, during the period December 2015 to September 2016. A total of 15 patients with prune belly syndrome were collected. Patients' characteristics were noted including socio-demographic data, laboratory and radiological investigations and any medical or surgical intervention. There were 12 males and 3 females with a male to female ratio of 4:1. Most of the patients (80%) had hydronephrosis and hydroureter. The study revealed that 60% of the patients had associated anomalies, there were 4 (26.6%) with cardiac defects, 3 (20%) with orthopaedic defects one patient with small bowel volvulus and one patient with cleft lip. 6 (40%) patients received medical intervention and 8 (53%) patients underwent surgical procedures. At the last follow up visit, 2 (13.4%) patients had normal renal function tests, 8 (53.3%) ended with chronic kidney disease, and 5 died with a mortality rate of 33.3%. Prune belly syndrome is a rare entity with wide variability in severity and clinical manifestations. The mortality in prune belly syndrome remains high despite medical and surgical interventions.

**Key words:**

Prune Belly Syndrome; Renal Anomaly; Undescended Testis; Abdominoplasty; Sudan.

## INTRODUCTION

Prune belly syndrome (PBS) (Triad syndrome, Eagle Barret syndrome) is a rare congenital anomaly characterized by a triad of deficient abdominal wall muscle, cryptorchidism and urinary tract anomalies, affecting 3.8 per 100,000 live male

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births [1,2]. Over 95% of patients are boys and only 3-5% are girls [3]. The cause of PBS is unknown, several aspects of the syndrome, including familial case reports and a higher incidence in males, suggest that it is influenced by a sex-linked genetic factor, although some have suggested an autosomal recessive mode of inheritance [4]. 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 [5]. Up to 75% of patients with PBS have associated pulmonary, skeletal, cardiac, and gastrointestinal defects [6]. There are few reports from developing countries regarding the pattern of renal involvement and management outcome of patients with PBS, but these reports are deficient due to lack of sufficient follow up to determine the course of the disease [7].

We report on 15 patients with PBS with wide variability in severity and clinical manifestations, most of the patients had associated anomalies and the majority had hydronephrosis and hydroureter. Despite medical and surgical interventions, the mortality remains high among this group of patients.

# METHODS

This was a prospective, case finding study that was conducted in the main paediatric hospitals in Khartoum state, the capital of Sudan. The study was conducted during the period December 2015 to September 2016. All children with a clinical and radiological diagnosis of prune belly syndrome or one of its variants, males and females, below 18 years of age, were included in the study. A total of 15 patients with PBS were collected. The diagnosis of PBS was usually evident at birth clinically. Patients with prune belly–like variant were also included in the study, these usually have abdominal wall defect without urological involvement. Patients were seen either as inpatients or in the outpatient clinics and examined by a senior Registrar or a Consultant Paediatrician.

A detailed questionnaire was filled out by the researchers, containing socio-demographic data, laboratory and radiological investigations and any medical or surgical intervention done for these patients. Dysmorphism and any associated anomalies were also looked for. The morphology of the urinary tract was assessed with ultrasonography and micturating cysto urethrography (MCUG). Diethylene Triamine Pentaacetic Acid (DTPA) renal scan was done for selected patients. Repeat investigations were individualized based on each case presentation and progress. Regular outpatient follow up was maintained for all cases during the nine months period of the study. Glomerular filtration rate (GFR) for children aged 1-18 years was calculated using the bedside Schwartz equation 2009 [8]:

Estimated glomerular filtration rate (eGFR) = 0.413 × (height/Scr)

Height is expressed in centimeters.

eGFR = mL/min/1.73 $m^2$

Standardized serum creatinine (Scr) = mg/dL

Stage of chronic kidney disease (CKD) was determined according to GFR as per the national kidney foundation guidelines [9]. For infants, less than one year of age we used reference values for serum creatinine in different subgroups, where 2.5th, 10th, 90th and 97.5th percentiles were the percentiles predicted from the model [10]. Blood pressure was measured using standard sphygmomanometer and cuff as well as a Dinamap monitor. Data was analyzed using Statistical Package for Social Sciences (SPSS) for windows version 16. Frequency analysis for background variables was conducted.

# RESULTS

The study group comprised 12 males and 3 females with a male to female ratio of 4:1. Regarding the age distribution, there were 3 patients less than one month of age, 7 patients between 1-5 years and 3 above 5 years of age (Table 1).

7 patients (46.7%) were diagnosed antenatally and the rest weren't. There were 14 patients with typical PBS and only one patient with prune belly like variant who had the typical absent abdominal wall muscle and normal renal ultrasound. Most of the patients (80%) had hydronephrosis and hydroureter, 13.3% had megacystis and one patient had a normal renal ultrasound (Table 2).

MCUG was done for 12 out of 15 patients, 66.6% of the patients had vesicoureteric reflux (VUR), one patient had patent urachus plus VUR and 13.3% had posterior urethral valve (PUV) in addition to VUR (Table 3).

Regarding the degree of VUR, one patient had grade 2, 2 patients (13.3%) had grade 3, 3 patients (20%) had grade 4 and 4 patients (26.7%) had grade 5.

When the associated anomalies were assessed, our series revealed that 60% of the patients had associated anomalies. There were 4 (26.6%) with cardiac defects, 3 (20%) with orthopaedic defects mainly talipes deformity and scoliosis, one patient with small bowel volvulus and one patient with cleft lip (Table 4).

DTPA scan was done for 4 patients out of 15 due to availability and affordability, 3 patients had severe renal damage on the left kidney and one patient had severe damage on the right kidney with poor drainage and mild to normal kidney function on the contralateral side. Regarding base line and follow up serum creatinine for patients less than one year of age, three of them had serum creatinine above the 97.5th percentile, one patient on the 90th percentile and one was below the 2.5th percentile. At last follow up for the same patients, three were above 97.5th percentile, one on the 90th percentile and one was between the 10th and 90th percentile. GFR at follow up visits for children above one year of age showed that there was one patient with normal GFR, one patient with end stage kidney disease (stage 5) and 8 patients with CKD. Table 5 illustrates the GFR and the corresponding stage of CKD in patients above one year of age.

With regards to the treatment given to these patients, 6 (40%) patients received medical intervention like prophylactic antibiotics, renal replacement therapy and mechanical ventilation. 8 (53%) patients underwent surgical procedures, about 50% of the patients had abdominoplasty and orchidopexy, one patient had vesicostomy and another one had cystoscopy with PUV ablation. Table 6 illustrates the surgical procedures done.

At the last follow up visit, 2 (13.4%) patients had normal RFT, 8 (53.3%) ended with CKD, and 5 died with a mortality rate of 33.3% among our series.

**Table 1- Age distribution of patients with prune belly syndrome**

| Age | Frequency | Percentage |
|---|---|---|
| **0-1 month** | 3 | 20.0 |
| **>1month - <1year** | 2 | 13.3 |
| **>1 - <5 years** | 7 | 46.7 |
| **> 5 year** | 3 | 20.0 |
| **Total** | 15 | 100.0 |

**Table 2- Ultrasound findings in patients with prune belly syndrome**

| Ultrasound findings | Number | Percentage |
|---|---|---|
| **Normal** | 1 | 6.7 |
| **megacystis only** | 2 | 13.3 |
| **Hydronephrosis + hydroureter** | 8 | 53.3 |
| **Hydronephrosis + hydroureter + dysplastic kidney** | 1 | 6.7 |
| **Hydronephrosis + hydroureter + megacystis** | 1 | 6.7 |
| **Hydronephrosis + hydrouriter + megacysitic + dysplastic kidney** | 2 | 13.3 |
| **Total** | 15 | 100 |

**Table 3- Micturating cystourethrogram findings in patients with prune belly syndrome**

| MCUG findings | Number | Percentage |
|---|---|---|
| **Not done** | 3 | 13.3 |
| **Normal** | 1 | 6.7 |
| **Megacystis only** | 1 | 6.7 |
| **Patent urachus + VUR** | 1 | 6.7 |
| **Megacystis + VUR** | 2 | 13.3 |
| **PUV + VUR** | 2 | 13.3 |
| **VUR** | 5 | 66.6 |
| **Total** | 15 | 100 |

MCUG - Micturating cystourethrogram, PUV - Posterior urethral valve, VUR - Vesicoureteric reflux

**Table 4- Associated anomalies in patients with prune belly syndrome**

| Anomalies | Number | Percentage |
|---|---|---|
| **Cardiac defect** | 4 | 26.6 |
| **ASD** | 3 | – |
| **PDA** | 1 | – |
| **Orthopaedic** | 3 | 20 |
| **Talipes equiovarus** | 1 | – |
| **Talipes calcaneoovalgum** | 1 | – |
| **Scoliosis** | 1 | – |
| **Gastrointestinal system** | 1 | 6.7 |
| **Small bowel volvulus** | 1 | – |
| **Others (cleft lip)** | 1 | 6.7 |
| **No associated anomalies** | 6 | 40 |
| **Total** | 15 | 100 |

ASD - Atrial septal defect, PDA - Patent ductus arteriosus

**Table 5- Glomerular filtration rate at follow visit for patients more than one year of age**

| Age at follow up | GFR (ml/min per 1.73m$^2$) | Stage | Comment |
|---|---|---|---|
| **1 year** | 30 | S III B | CKD |
| **16 months** | 47 | S III A | CKD |
| **2 years** | 65 | S II | CKD |
| **2.25 years** | 28 | S IV | CKD |
| **2.50 years** | 68 | S II | CKD |
| **2.50 years** | 118 | S I | Normal |
| **3 years** | 41 | S III B | CKD |
| **6 years** | 84.7 | S II | CKD |
| **7 years** | 24 | S IV | CKD |
| **10 years** | 6.7 | S V | ESKD |
| **Total (n = 10 patients)** | – | – | 66.7% |

CKD - Chronic Kidney disease, ESKD - End stage kidney disease, GFR - Glomerular filtration rate

**Table 6- Surgical intervention for patients with prune belly syndrome**

| Intervention | Number | Percentage |
|---|---|---|
| **Cystoabdominoplasty and orchidopexy** | 1 | 6.7 |
| **Abdominoplasty and orchidopexy** | 2 | 13.4 |
| **Orchidopexy** | 1 | 6.7 |
| **Vescicostomy** | 1 | 6.7 |
| **Gastrojejunostomy** | 1 | 6.7 |
| **Cystoscopy with PUV ablation** | 1 | 6.7 |
| **Others** | 1 | 6.7 |
| **No intervention** | 7 | 46.6 |
| **Total** | 15 | 100 |

# DISCUSSION

Prune Belly syndrome (PBS) was first described by Frolich in 1839 and it is a complex malformation disorder with wide variability in severity and clinical manifestations [11]. Though rare, cases of familial PBS have been reported [4]. None of our patients however has any positive family history.

Our series revealed that male to female ratio in children with PBS was 4:1 which indicates a higher female preponderance than what is reported in the literature, this is hard to explain but as this is a case finding study, some cases might have been missed. The PBS is 20 times more common in males than female and this has been attributed to the more complex morphogenesis in males than in females (Figure 1) [12, 13].

The present study showed that only 46.7% of patients with PBS were diagnosed antenatally, which might indicate either poor antenatal care attendance or lack of accuracy of antenatal ultrasonography. Prenatal diagnosis plays a key role in early detection of PBS, with prenatal ultrasonography; PBS can usually be diagnosed in the second trimester of pregnancy [14].

Our series revealed that 60% of the patients had associated anomalies, which is compatible with what is reported in the literature [6]. 26.6% of patients with PBS had cardiac defects mainly atrial septal defects; this is higher than 10% reported cardiovascular involvement [15]. Given this association, it is recommended that all prune belly patients should undergo cardiovascular evaluation. Our series showed that 6.7% of the patients had associated gastrointestinal anomalies mainly small bowel volvulus, this is lower than what is reported in the literature of 20-30% which is thought to result from an insufficient fixation of the mesentery to the back of the abdominal wall [16, 17]. Twenty percent of the patients in our series had orthopaedic defects mainly talipes deformity and scoliosis, which is lower than 45-63% reported associated orthopaedic defects [18], More severe deformities of the musculoskeletal system such as limb deformities and arthrogryposis appear to be secondary to oligohydramnios causing compression of the developing fetus [19].

The current case series showed that most of the patients (80%) had hydronephrosis and hydroureter, which is similar to other reports, as hydronephrosis is a common finding and is usually detected during antenatal care visits [20]. About 50% of the patients in our series had abdominoplasty and orchidopexy. In addition to the cosmetic effect of abdominoplasty, there is an additional advantage of improvement in bladder emptying with a marked reduction in post void residual urine and improved bowel function (Figure 2) [21]. It is recommended that orchidopexy should be performed as early as possible since the endocrine function is preserved and there is a malignant potential [22].

From our series, the overall mortality rate of 33.3% compares with the worldwide mortality rate of 20-30%, mortality in PBS is usually due to pulmonary hypoplasia and renal failure [7]. The prognosis in PBS is variable and depends on the degree of pulmonary hypoplasia, associated renal anomalies, and degree of renal compromise [23].

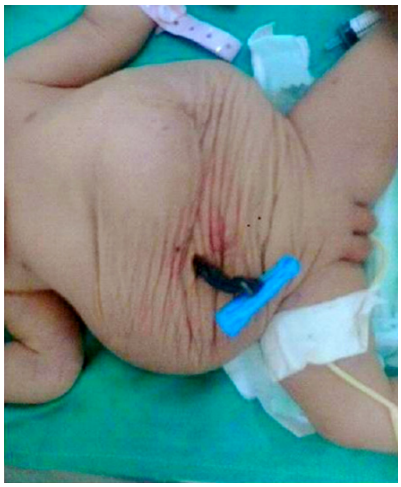

**Figure 1- Female baby with Prune belly syndrome**

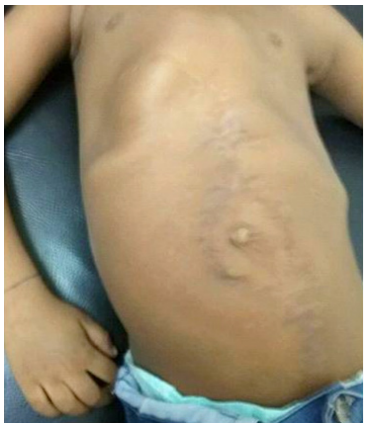

**Figure 2- A child with Prune belly syndrome who had abdominoplasty**

## CONCLUSION

PBS is a rare entity worldwide with wide variability in severity and clinical manifestations, most of the patients had associated anomalies and the majority had hydronephrosis and hydroureter. The mortality in PBS remains high despite medical and surgical interventions. Prenatal diagnosis plays a key role in early detection of PBS.

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