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ORIGINAL ARTICLE

PRUNE BELLY SYNDROME; A COMPLEX CONGENITAL ANOMALY OF 10 CASES FROM A TERTIARY HOSPITAL

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ABSTRACT

Introduction*: A complex congenital anomaly is a rare congenital multisystemic disorder characterized by absence of abdominal wall muscles, dilated urinary bladder, a variety of other urologic abnormalities and undescended testes in males. Patients could have pulmonary hypoplasia, a variety of cardiac abnormalities and kyphoscoliosis. Here we present 10 pediatric patients who were on follow up at our renal clinic of the Department of Pediatrics and Child Health of Tikur Anbessa Specialized Hospital.*

Patients and methods*: A prospective case finding study was conducted in the renal follow up clinic of the Department of Pediatrics and Child Health of Tikur Anbessa Specialized Hospital from January 2019 to January 2020.*

Results*: There were ten children on follow up. Their age ranges between three months to ten years. Age at first diagnosis ranges from 7 days to 9 years. All were males, had undescended testis, hydronephrosis, hydroureters and megacystis. Three patients had stage 5 chronic kidney disease and two patients died.*

Conclusion*: Though there is no consensus to the management of Prune Belly Syndrome; individualized medical and surgical management would improve survival than a wait-and-see approach. Early diagnosis before deterioration of renal function is important.*

Key words*: Prune Belly Syndrome, cryptorchidism, hydronephrosis, kyphoscoliosis*

INTRODUCTION

Prune Belly Syndrome (PBS) is a rare congenital disorder characterized by hypoplasia of the abdominal wall muscles, bilateral undescended testes and malformation of the urinary tract (1-3). There could also be associated pulmonary hypoplasia, cardiac abnormalities, skeletal malformations and gastrointestinal abnormalities (4, 5). It affects one in 30,000-40,000 live births almost all are males and 3-5% females (3, 4).

Currently prenatal diagnosis is possible through antenatal ultrasound examination in the second trimester of pregnancy. Oligohydramnios, lax abdominal wall, various degrees of hydronephrosis, renal dysplasia could be observed; in severe forms the fetus might not be viable (6, 7).

Prune belly Syndrome was first described in 1839 by Frölich, and the syndromic name was coined in 1901 by Osler due to a dried prune like abdominal wall resulting from absence of the abdominal wall muscles (8, 9).

In this study, 10 cases of Prune Belly Syndrome with a variety of clinical manifestations, course of the disease and the management challenges were reported.

PATIENTS AND METHODS

A prospective case finding study was conducted in the renal follow up clinic of the Department of Pediatrics and Child Health of Tikur Anbessa Specialized Hospital from January 2019 to January 2020. Of the ten children who were on regular follow up with the diagnosis of Prune Belly Syndrome (PBS) during and after the study period, two died during the past one year.

Detailed information was taken using a questionnaire including demographic characteristics, anthropometric assessment, investigations (including imaging, blood chemistry, and urinalysis). Thorough physical examination was done, and additional investigations were performed based on the clinical findings. Ultrasound examination was done for all cases; voiding cystourethrography was done when service was available and echo-cardiography was done if there were indications. Radionuclide renal scan was not, however, done because of unavailability of the service in the hospital. Repeated investigations were done when necessary.

Glomerular filtration rate was calculated for children between 1-10 years using the modified Schwartz formula (11): Estimated glomerular filtration rate (eGFR) = 0.413 × (height/length in cm/serum creatinine in mg/dl) = ml/minute/1.73m^2.

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We determined the stage of chronic kidney disease following the guidelines of the National Kidney disease foundation (stage 1-5 based on the level of eGFR) (12).

For infants less than 12 months of age, we used reference values for serum creatinine; where 2.5th, 10th, 90th and 97.5th percentiles were the reference ranges according to Boer DP. *et al*. (13).

Ethical clearance: Care takers consented verbally and the study was cleared ethically by the research and publication committee of the department.

RESULTS

There were ten study children and all were males. Their age ranged from 3 months to 10 years and all had cryptorchidism. The age distribution is as shown in table 1 and figures 1-2. Age at first diagnosis ranged from 7 days to 9 years.

Third trimester ultrasound showed fetal hydronephrosis in 2 patients. There was no associated cardiac defect, pulmonary hypoplasia, gastrointestinal abnormality like volvulus or cleft lip. A one year and five-month-old patient had deformed chest with kyphoscoliosis, a three-month old-child had craniosynostosis. The other eight patients did not have orthopedic abnormalities. All had megacystis, hydroureters and hydronephrosis. Ultrasound examination findings are shown in table 2.

Table 1: Age distribution of patients with Prune Belly Syndrome

Age	Frequency	Percentage
Months	2	2/10 (20)
>3 months -1year	2	2/10 (20)
>1year -2 years	2	2/10 (20)
>2years -5years	1	1/10 (10)
>5years-10 years	3	3/10 (30)

Figure 1: A 3-month-old male child with PBS

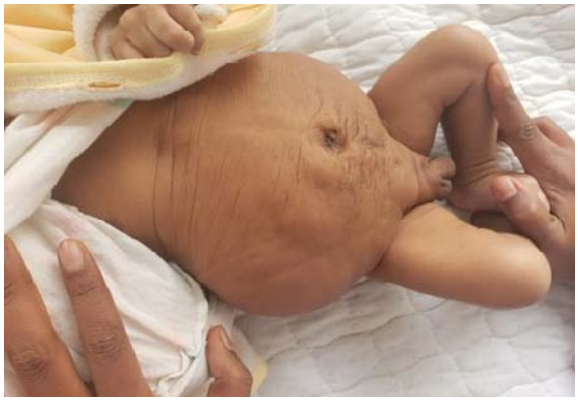

Figure2: A 7- month- old male child with PBS

Table 2: Ultrasound findings of children with Prune Belly Syndrome

Ultrasound findings	Number	Percent
Hydronephrosis, hydroureters, megacystis	10	10/10 (100)
Right side multicystic hypoplastic kidney	1	1/10 (10)
Dysplastic kidney (right)	1	1/10 (10)
Bladder diverticula	1	1/10 (10)
Urachal diverticula	1	1/10 (10)

Voiding cystourethrography was done for 6 patients, and there was huge dilated bladder in all of them, vesico-ureteral reflux grade 4-5, hydroureters and hydronephrosis. Four children had dilated prostatic urethra, and posterior urethral valve was considered as shown in table 3.

The stage of kidney disease was assessed in patients above one year of age based on the eGFR. Three patients had stage 5 CKD with eGFR less than 15 ml; three patients had stage 3 CKD with eGFR between 30-60 ml and one patient had stage 1 CKD with eGFR between 90 and 120 ml. Three infants less than one year of age had normal renal function. On the last day of their visit, 17 months and a 10-year-old child died due to end stage renal disease.

Concerning surgical interventions, orchidopexy was done in one patient, while r orchidectomy was done in another patient; no other surgical intervention was done. All eight patients were on supportive medical treatment for chronic kidney disease and on prophylaxis for urinary tract infection.

Table 3: findings of voiding cystourethrography in children with Prune Belly Syndrome

VCUG findings	Number	Percent
Dilated prostatic urethra, posterior urethral valve, grade 5 VUR (left), no reflux (right), dilated bladder, bilateral severe hydroureteronephrosis	1	1/10 (10)
Dilated and thickened bladder, bilateral grade 4 VUR, severe hydroureteronephrosis	1	1/10 (10)
Dilated bladder, bilateral grade 5 VUR, hydroureter-onephrosis	1	1/10 (10)
Grossly dilated bladder and prostatic urethra, posterior urethral valve and grade 5 VUR bilateral hydroureter-onephrosis	2	2/10 (20)
Huge bladder with diverticula, dilated prostatic urethra, posterior urethral valve grade 4 VUR	1	1/10 (10)

VUR-vesico-ureteral reflux

DISCUSSION

PBS occurs often in males, and there is variability in severity ranging from fatality at birth to long years of survival with surgical intervention (14). All patients were males, the current survival ranged from infancy to 10 years. The eldest survivor was ten years old. Familial predisposition in twin pregnancies was described in some literature (4, 10, 15), but there was no familial predisposition in our cases; no consanguineous marriage and all were singleton deliveries.

At 11-12 weeks of gestation antenatal diagnosis of oligohydramnios and hydronephrosis was possible though detailed description made at second trimester of pregnancy. In our study antenatal hydronephrosis was detected in only 2/10 (20%) patients which was much lower than other studies (16). This might be because of poor antenatal care or unavailability of ultrasound machine. Regarding other congenital anomalies, one child had chest wall deformity and kyphoscoliosis and another child had craniosynostosis. No other associated congenital anomaly was detected which is an indicator of a better prognosis if appropriate surgical intervention is made with supportive medical treatment. This finding is similar to other series with variability of severity of manifestations (4, 14).Follow up ultrasound scan showed that all of our patients had hydronephrosis, hydroureters and dilated bladder which is in agreement with the results of other studies (4, 17). Voiding cystourethrography done in 6 patients showed grade 4-5 vesico-ureteral reflux, while the one done in four patients dilated prostatic urethra and posterior urethral valves.

This finding is I agreement with that of a study done in the Sudan (4) though the number of children with posterior urethral valves is higher in our study.

Regarding the stage of chronic kidney disease in children above one year of age; 5/10 (50%) had CKD among which 2/10 (20%) died. Mortality rate which is 20-30% is comparable to other studies (18) but ten-year survival rate was much lower in our case series than others (17) probably due to absence of surgical intervention. Mortality was due to end stage renal disease.

Supportive medical treatment for possible recurrent urinary tract infection and electrolyte abnormalities was given. Management was individualized depending on the severity of urological malformations and other co-morbid systemic abnormalities. These abnormalities also determine survival rate in PBS. Staged and individualized surgical intervention with medical management would improve survival since there is no consensus reached guideline for the management of these cases (17).

Conclusions

Though there is no consensus in the management of PBS; individualized medical and surgical management would improve survival than a wait-and-see approach. Early diagnosis before deterioration of renal function is important.

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