

Prune-Belly syndrome in 2 cases seen in a tertiary medical institution Southeast Nigeria – A case report

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ABSTRACT

Prune-Belly syndrome is a rare congenital disorder, and in underdeveloped and developing countries, the outcome is not well known as only a few isolated cases have been reported. A review of 2 male neonates admitted and managed for Prune-Belly syndrome in the neonatal unit of the Pediatric Department of the Federal Teaching Hospital of Nigeria. This appears to be the very first of such cases to be seen at this hospital. Both babies were delivered outside this tertiary institution and eventually referred here for proper diagnosis and management. Two male neonates aged 2 and 3 days, respectively, were admitted in the same month with a common history of maternal febrile history, poor cry on delivery, and oligohydramnios in one of them. Clinical examination showed scaphoid, lax, and wrinkled abdomen with visible peristalsis and flank fullness, ballotable kidneys, distended bladder, well-formed phallus, small scrotum, and absent testes. There were also musculoskeletal abnormalities ranging from lower limb dysgenesis to clubbing of the feet. Prune-Belly syndrome presents with a spectrum of features which present an overwhelming challenge to both the parents and the managing physician, especially in most resource-poor countries.

Key words: *Musculoskeletal abnormalities, Neonates, Prune-Belly syndrome, Underdeveloped countries*

Pruno-Belly syndrome (PBS) (Triad syndrome, Obrinsky syndrome, or Eagle-Barrett syndrome) is a rare congenital anomaly characterized by a trinity of deficient abdominal wall muscle, bilateral cryptorchidism, and urinary tract anomalies which may include megaureter, hydroureter, hydronephrosis, vesicoureteral reflux, and megacystis. Pulmonary, orthopedic, cardiac, and gastrointestinal abnormalities may also be present [1-8].

The incidence of occurrence is 1/30,000–40,000 live births, affecting mostly males with only 3–4% being females and 4% of all cases being products of twin pregnancies [1-4]. PBS has no established etiology. However, it is thought to be associated with trisomy 18 and 21 and seems to occur more in some congenital cardiovascular disorders including tetralogy of Fallot and ventricular septal defects [7]. There is no known underlying genetic cause although a recent mutation in the Cholinergic Receptor Muscarinic 3 gene has been reported in one family with a history of PBS [1,2,8]. The deletion of hepatocyte nuclear factor-1 beta, a transcription factor required for visceral endoderm specification during embryogenesis by recent findings, increasingly also supports a genetic background in PBS patients [8-11].

Infant mortality is decreasing worldwide; however, neonatal mortality is still high owing to the cases of congenital anomalies,

including those arising from complicated or poorly managed PBS cases [12]. It is a known fact that limited resources for permanent renal replacement therapy are a potential factor in increased mortality rates in such conditions when they are compounded with chronic kidney disease and end-stage kidney disease in our setting. We are reporting two cases which presented with typical features of PBS and were delivered outside the facility. This report is expected to raise the awareness among the medical doctors and other health-care professionals. It is also expected that it would help give emphasis to the need to include prenatal ultrasound scanning for all pregnant mothers such that suspected cases are handled from the beginning by the experts in the field.

CASE REPORT

The first baby was a male admitted to the Special Care Baby Unit (SCBU) of Federal Teaching Hospital Abakaliki (FETHA) on the 3rd day of life with poor cry at birth, refusal to suck, abnormally shaped abdomen, and inability to pass urine since birth (Figs. 1 and 2) and delivered at a private hospital and transferred to the tertiary institution. Mother was a 32-year-old primiparous unemployed, school certificate holder, married to a 35-year-old patent medicine vendor with a primary school



Figure 1: The first baby with the wrinkled and malformed anterior abdominal wall with scissoring of the lower limbs



Figure 2: Side view of the first baby shown with the wrinkled and malformed anterior abdominal wall with scissoring of the lower limbs

certificate. She had febrile illnesses without any skin rash at the 5th and 7th month of gestation, respectively, which resolved on treatment with oral antimalarial drugs. Prenatal ultrasound scan demonstrated the baby's large abdominal without documentation of the liquor volume.

The baby weighed 3.4 kg, noted to be in respiratory distress, febrile, dehydrated, and mildly pale. The abdomen was distended with visible peristalsis, distended anterior abdominal wall veins wrinkled with flank fullness, partial absence of the abdominal muscles, and a patent anus. Kidneys were ballotable with a distended bladder, well-formed phallus, and small scrotum with absent testes. The baby had pectus excavatum, tachypneic (respiratory rate of 66 breaths/min) with clear lung fields, was fully conscious, and had widened sagittal suture as well as patent anterior and posterior fontanelles. There was increased muscle tone with scissoring of the lower limbs while the upper limbs had normal tone.

The second baby, a 2-day-old term male neonate, was admitted in the SCBU of FETHA with complaints of fast breathing, abnormal-shaped abdomen, abnormal-shaped lower limb, and inability to pass urine since birth (Figs. 3 and 4). He



Figure 3: The second baby with dysgenesis of the right lower limb and wrinkled malformed anterior abdominal wall



Figure 4: The second baby with the dysgenesis of the right lower limb and wrinkled malformed anterior abdominal wall

was delivered outside the hospital at a gestational age of 40 weeks through spontaneous vertex delivery. Mother was a 38-year-old grand multiparous petty trader married to a 48-year-old mason. The baby was the 5th child in a monogamous setting with all the siblings alive and well.

Mother had a febrile illness without any skin rash at the 2nd month of gestation which resolved following treatment with some unknown drugs. However, ultrasound scan was not done during pregnancy though history showed a poorly growing abdominal girth that was non-commensurate with the gestational age, suggesting a possible oligohydramnios. By the 6th month of gestation, she was admitted for threatened abortion.

The baby weighed 2.4 kg, hypothermic (T-34.6°C), lethargic, was in respiratory distress with a respiratory rate of 80/min, and had a short and narrow chest cavity with normal auscultatory findings. The abdomen was scaphoid, lax, and wrinkled with visible peristalsis, flank fullness, and absence of the abdominal muscles but had a patent anus. Kidneys were ballotable with a distended bladder (up to the umbilicus and firm). The phallus was well formed but with an empty small scrotum. The baby had a noticeable right lower limb dysgenesis, and left lower limb clubbing though the tone was normal.

Both sets of parents were counseled about the diagnosis and prognosis of the condition. Investigations including urinalysis with microscopy, culture and sensitivity, complete blood count, chest X-ray, abdominopelvic ultrasound and serum electrolyte, urea, and creatinine were requested, and the results are as shown in Tables 1 and 2.

The renal function test demonstrates a progressively rising serum urea and creatinine and a falling sodium rate. Urinalysis showed pH of 8.0, blood +3, and glucose +1 without any pus cells or bacteria. Culture was not done due to the financial constraints. Complete blood count showed packed cell volume (PCV) of 32 % and white blood cell (WBC) of 19600/mm³. Abdominal ultrasound showed enlarged kidneys with marked dilatation of the pelvicalyceal system, ballooning of calyces, and thickening of the renal cortex; the cortices of both kidneys show tiny cystic spaces. Right kidney measured 6.9 cm × 2.8 cm while the left kidney measured 6.0 cm × 3.0 cm. Both ureters were markedly dilated and tortuous along their full length, and the urinary bladder was mildly filled with urine and thickened with irregular mucosa housing multiple floating echogenic masses. The anterior wall of the abdomen was thin, with thin muscle slips. The scrotal sacs were empty as testes could not be demonstrated in the groin or abdominal cavity.

As shown in table, the renal function tests showed a gradually decreasing serum urea, creatinine and sodium rate, and a high potassium levels. Urinalysis showed blood +3 but urine culture showed no growth. PCV was 30% on presentation and eventually decreased to 18% by the 31st day on admission, and total WBC was 15,200/mm³. Catheter tip culture yielded *Enterobacter* spp. sensitive to only ofloxacin. Ultrasound showed bilateral hydronephrosis, hydroureter with evidence of bladder outlet obstruction. The anterior wall of the abdomen was thin, with empty scrotal sacs (no testes).

The babies were commenced on intranasal oxygen, intravenous fluids, and antibiotics, nasogastric tube feeding with expressed breast milk. Other specialties including the pediatric surgeons,

urologist, radiologist, orthopedic surgeons, and the social workers were invited to review and comanage the babies.

All silicone-coated urinary catheters were passed for continuous bladder drainage in both cases which drained bloody urine for several days, clearing up from the 9th day on admission for the 1st baby while the urine was almost clear for the 2nd baby. Urine output ranged from 0 to less than 20 ml/day while it ranged from 1 ml/kg/h to 3 ml/kg/h for the 2nd baby. Correction for dyselektrolytemia was instituted.

Condition of the first baby continued to deteriorate despite treatment. Plans for urine diversion (ureterostomy) were made; however, parents were unwilling to consent because of financial constraints but signed against medical advice on the 11th day on admission. Baby has been lost to follow-up since on discharge.

The 2nd baby developed a high-grade fever with purulent urinary discharge. Urinary catheter tip culture yielded *Enterobacter* spp. sensitive to only ofloxacin, which was commenced immediately. The pediatric surgeon discussed with the parents on the need for a urinary diversion (nephrostomy) which again was rejected by the parents due to financial constraints as well as hopelessness of the baby's condition. By the 38th day on admission, baby died from cardiopulmonary failure secondary to overwhelming septicemia.

DISCUSSION

PBS is a congenital disorder characterized by the clinical triad of cryptorchidism, hypoplasia of the abdominal musculature, and abnormalities of the urinary tract. The term "Prune Belly" reflects the characteristic wrinkled appearance of the abdominal wall in the newborn due to the complete or partially complete absence of abdominal wall muscles [1-4]. It has no established etiology though some cases of familial PBS have been reported [13,14]; however, in our index cases, none of the babies had any positive family history of a similar anomaly.

Ninety-five percent of cases occur in males (as was seen in our study, all were males) with the diagnosis made in utero using ultrasound scan usually suspected when imaging reveals an enlarged bladder, dilated ureters, and an abnormal abdominal wall. This was partially noted in the first baby who had a prenatal ultrasonographic scan which revealed an enlarged abdomen even though details of the size of the renal tract components were not discussed.

According to earlier studies, PBS is associated with age [15] and it occurs more among babies of young mothers. However,

Table 1: Renal function test results for the first baby

Renal function test	1 st day on admission	5 th day on admission	10 th day of admission
Urea (mmol/l)	22.4	39.2	74
Creatinine (umol/l)	481	730	876
Sodium (mmol/l)	145	131	126
Potassium (mmol/l)	4.5	4.9	4.6
Chloride (mmol/l)	110	96	78

Table 2: Renal function test results for the second baby

Renal function test	1 st day	After 3 days	After 7 days	After 10 days	After 21 days	After 25 days
Urea (mmol/l)	23.0	28.3	25.5	31.1	27.1	22.1
Creatinine (umol/l)	457	483	428.7	472	439.8	319.6
Sodium (mmol/l)	135	126	118		122	119.2
Potassium (mmol/l)	6.7	7.8	5.1		6	6
Chloride (mmol/l)	98	93	91		94	93.3
Bicarbonate	-	-	-	-	-	8.5
Anion gap	-	-	-	-	-	23.4

this was not the case in our patients whose mothers were 32 and 38 years, respectively. There was a history a febrile illness in both mothers during pregnancy while at different gestational periods although with no confirmed etiology.

There are other associated anomalies, including clubfoot, chest wall deformities such as pectus carinatum, intestinal malrotation, pulmonary dysplasia, congenital hip dislocation, and cardiac defects due to the sequela of oligohydramnios in utero. Some of these musculoskeletal abnormalities were observed in our index patients [1-8].

Morbidity is associated with the degree of dilation of the urinary tract which results in poor ureteral peristalsis and weak forward propulsion of urine in the ureters. This, in turn, results in stasis, infection, and stone formation. Renal failure which may be demonstrated by the varying degrees of electrolyte impairment (dyselectrolytemia) often results from underlying renal dysplasia and the aforementioned complications of urinary stasis [1-8]. The two cases reported in the current study also had impaired renal function with dyselectrolytemia. Virtually, all males have bilateral undescended testes – cryptorchidism. Females usually have normal ovaries but abnormalities of the uterus and vagina. The empty scrotal sacs (cryptorchidism) were equally observed in our study patients.

In most resource-poor sub-Saharan African countries, management of this condition is burdensome and difficult because of poor local resources for adequate management of renal failure including renal replacement therapy [16]. In addition, superstitious beliefs also confound the management and both resulting in limitation of adequate management as most families could seek discharge against medical advice with a loss to follow-up of cases [17], and mortality as was seen in the current report.

CONCLUSION

PBS manifests with a catalog of features. This condition often presents an overwhelming challenge to both the parents and the managing physician in most resource-poor countries.

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