

CASE REPORT

Prune Belly Syndrome in a Nigerian infant: a case of inevitable or preventable death?

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ABSTRACT

Background: Prune Belly Syndrome is a rare congenital disorder comprising deficient anterior abdominal musculature, undescended testes and urinary tract abnormalities. In Nigeria, the incidence is unknown but majority of the few reported cases were seen in the South west region of the country. Despite its poor prognosis, a coordinated multidisciplinary management may prevent early mortality.

Case Report: We report a 6-week old male infant brought to our health facility with the complaints of abdominal distension and abnormal feet since birth. Physical examination revealed a lax anterior abdominal wall, undescended testes, as well as bilateral *talipes equinovarus*. Radiologic evaluation showed urinary tract abnormalities such as dilated left ureter and pelvis and right multi-cystic, dysplastic kidney. The patient was treated with the appropriate antibiotic for culture-proven urinary tract infection after which he was referred to the Paediatric Surgeons. Few weeks after his discharge by the Paediatric Surgeons for subsequent follow-up in their clinic, the patient's sudden demise at home was reported to us.

Conclusion: The sudden mortality of this infant has brought to the fore the gaps in treatment which could have affected his longevity. This report underscores the need for a coordinated multidisciplinary approach in the management of this rare congenital anomaly in order to improve survival outcome in a resource-limited setting.

Keywords: Congenital anomaly, multidisciplinary management, prognosis, survival

INTRODUCTION

Prune Belly Syndrome is a rare congenital disorder comprising deficient anterior abdominal musculature, undescended testes and urinary tract abnormalities especially megacystis-megaureters, vesico-ureteric reflux and renal dysplasia.¹

It was first described by Froehlich in 1839 but Osler gave the condition its name.²The synonyms include Triad syndrome, Eagle-Barrett syndrome and Obrinsky syndrome. It

occurs almost exclusively in males and rarely in females. Its coexistence with limb, cardiopulmonary and gastrointestinal malformations is well documented.³ Reports also indicate its association with chromosomal abnormalities such as trisomy-21 and trisomy-18, as well as Beckwith-Weidman syndrome and VACTERL anomaly.^{4,6,7,8,9}

Approximately 30% of infants with Prune Belly Syndrome are still-born as survival

depends on the degree of pulmonary hypoplasia and renal dysplasia.³ Up to 30% of the long-term survivors may develop end-stage renal disease from dysplasia, reflux nephropathy or complications of infection.³

The world-wide incidence is 1:30,000 or 1:40,000 live births.² In Nigeria, the exact incidence is unknown probably due to scanty data from under reporting or missed diagnosis. Most of the published reports about this rare congenital anomaly emanate from the South-West Zone of the country.^{6, 10, 11, 12, 13, 14} Two reports have however been documented in South-East Nigeria.^{15, 16}

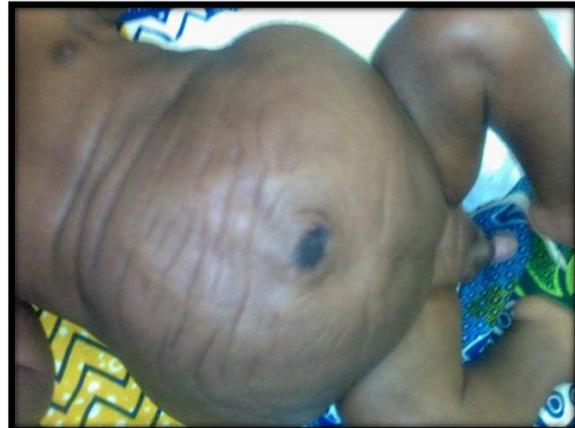
This case report was prompted by the need to compare the observations from the index patient with those from previous reports in Nigeria, as well as to highlight the importance of a coordinated multi-disciplinary management which may result in better survival outcome.

CASE REPORT

A 6-week old male infant was brought to the Paediatric Nephrology Clinic of the University of Nigeria Teaching Hospital, Enugu with the complaints of abdominal distension and abnormal feet since birth. There was no history of vomiting, change in bowel habit or urinary symptoms. The child's feet were noted to be curved inwards and upwards. Pregnancy history revealed decreased fundal height and reduced fetal movement compared to previous pregnancies. The index patient was the last in the family with 4 children. Other siblings were well. Mother was a 38-year old house wife and father was a 50-year old artisan (Electrician).

Physical evaluation showed an afebrile (temperature 36.5°C) male infant with no pallor, icterus or cyanosis. He was in mild respiratory distress with nasal flare,

Figure 1. A close up view of the patient showing a distended abdomen with wrinkled anterior abdominal wall



intercostal and subcostal recessions, although the respiratory rate was 42 breaths/minute. The breath sounds were unremarkable. The cardiovascular system was essentially normal. His length and weight were 48cm and 3.85 kg respectively. The abdomen was distended with lax anterior abdominal wall with visible peristalsis. The abdominal girth measured 42cm at a point 8cm from the xiphisternum in the midline. There was hepatomegaly of 4cm; firm, smooth and non-tender and a supra-pubic mass of about 12 weeks size. The right kidney was ballotable. The scrotum had normal rugae but both testicles were non palpable. He had a normal phallus, as well as bilateral *talipes equinovarus* (see Figures 1 and 2).

Laboratory evaluation included serum electrolyte, urea and creatinine estimation which showed sodium 139mmol/L (135-145mmol/L), potassium 4.2mmol/L (3.5-5.0mmol/L), bicarbonate 24mmol/L (24-32mmol/L), chloride 98mmol/L (97-108mmol/L), urea 3.5mmol/L (1.8mmol/L) and creatinine 42micromol/L (18-35micromol/L). The estimated GFR was 44.4mls/min/1.73m² (normal for age).

Figure 2. Another view of the same patient during hospitalization with obvious lax anterior abdominal wall



Full blood Count revealed red blood cell count $3.2 \times 10^6/L$, haemoglobin concentration-11.0g/dL, packed cell volume 33%, mean corpuscular haemoglobin concentration-33.2g/dl, mean corpuscular volume 101.8fl, platelet count $401,000 \times 10^9/L$, white blood cell count $11,700 \times 10^9/L$ (Neutrophils 36%, Lymphocytes 64%). Urine culture yielded staphylococcus aureus ($>10^5$ organisms/ml) sensitive to ceftriaxone, gentamycin and nitrofurantoin.

The imaging studies on the patient consisted of abdominal ultrasonography, intravenous urography, micturating cysto-urethrography and chest radiograph. The results of the chest radiograph and micturating cysto-urethrography could not be retrieved before this report. Abdominal ultrasonography revealed that the right kidney measured 6.2cm x 2.9cm showing as a multi-cystic mass with intervening septae. It extended across the midline to the left and inferiorly into the right iliac fossa. The left kidney was also visualized with a dilated renal pelvis and proximal ureter. The bladder wall thickness was 6mm (too thick for the child). The scrotal sacs were empty but the testes were not sonographically detectable even within the inguinal canal. The rest of the abdominal organs were unremarkable. The conclusions

made were left ureterectasis and pelviectasis, right multi-cystic dysplastic kidney, and thickened bladder wall; which were in keeping with Prune Belly Syndrome. The intravenous urography results from the contrast films showed prompt excretion by the left kidney with normal calyces but with full renal pelvis and dilated, redundant ureter. There was no excretory function demonstrated in the right kidney. The bladder was normal sized but mildly deviated to the left.

After a course of parenteral ceftriaxone to eradicate the culture-proven urinary tract infection, the patient was referred to the Paediatric Surgery Unit following clinical stabilization. From the Paediatric Surgery ward, he was discharged to the unit's outpatient clinic for subsequent follow up. Before he reached his 3rd month of life, the parents communicated to us about his sudden demise at home.

DISCUSSION

This third documented case of Prune Belly Syndrome in south east Nigeria presented with epidemiologic and clinical features similar to those of previous reports in the country.^{6,10,15} For instance, the male gender of our patient, the age of presentation, the advanced maternal age and the associated limb deformity (club foot or *talipes equinovarus*) are in keeping with the observations by Okeniyi, *et al*, as well as Adekanmi, *et al* who however noted a female phenotype in one of the three cases they reported.^{6,10} Other case reports nevertheless suggest that babies born with the syndrome were products of young maternal age.^{17,18} While the index case presented with urinary tract abnormalities such as multi-cystic dysplastic kidney and ureterectasis/pelviectasis, the report by Njeze and Okoye documented fourth degree reflux and mega ureters.¹⁵

The exact aetiology and pathogenesis of this disorder remain unclear and controversial.

Hitherto, previous findings suggest a functional urethral obstruction secondary to prostatic hypoplasia as the underlying cause of the components of the syndrome.^{19,20,21} The current prevailing theory however links Prune Belly Syndrome to a mesodermal arrest during embryogenesis as a result of exposure to a noxious insult during early period of the first trimester.² This latter hypothesis better explains the simultaneous involvement of the genitourinary tract, the testes and the anterior abdominal wall, as well as other associated malformations.

Although our report did not establish such exposure in the prenatal history of the index patient, other authors noted maternal ingestion of native herbal concoctions in their case reports.¹⁰ In a developing country such as Nigeria where prenatal diagnosis is not yet well developed, the importance of well supervised antenatal care services- in order to reduce maternal exposure to teratogens- cannot be over-emphasized.

Similar to our experience, most of the reported cases of Prune Belly Syndrome from south west Nigeria rarely survived beyond infancy.^{6,10,14} However, Salako and his colleagues had documented a 24-year old male adult presenting with this anomaly.¹³ Although the current mortality rate is put at 20%, the patient's survival depends on the severity of co-morbidities and their sequelae.²

The early mortality in the index patient (who was discharged home in a stable clinical state) poses a fundamental question about his survival: was his death inevitable or preventable? Obviously, a coordinated multi-disciplinary approach and collaboration among Paediatric Nephrologists, Paediatric Surgeons, Urologists, Plastic and Orthopaedic Surgeons, as well as paramedics could have helped to improve the prognosis and quality of life. This initial collaboration to formulate and prioritize the treatment options was admittedly missing.

The patient would have benefitted from periodic monitoring of renal function and antibiotic prophylaxis to prevent recurrent urinary tract infection, since as many as 30% of long-term survivors develop end-stage renal disease from dysplasia or reflux-related/infection-related complications.³ Advances in renal replacement therapy especially in developed countries, as well as surgical interventions have indeed led to favourable outcome.³

The exact cause of death in this patient, nevertheless, remains conjectural. If it was due to pulmonary complications, it was inevitable since mortality in 30% of affected infants in the first few months is attributed to pulmonary hypoplasia.³

However, prognosis depends on the degree of pulmonary hypoplasia, as well as renal dysplasia. Our patient did not appear to have had severe pulmonary hypoplasia. Better still, he showed evidence of a functional left kidney in spite of a right multi-cystic dysplastic kidney; which underscores our presumption that his sudden death at home could have been preventable if he was closely monitored and followed up.

CONCLUSION

This case report has highlighted the need for a coordinated, multi-disciplinary approach in the management of patients with this rare congenital anomaly. This may improve their quality of life and the chances of survival beyond infancy.

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