A First Description of Prune Belly Syndrome in Central Africa

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1. Introduction

Prune belly syndrome (PBS) is a rare congenital disease that was first described by Frohlich in 1839, and the name PBS was coined by Osler in 1901. PBS is characterized by deficient abdominal wall musculature, hypotonia, urinary tract dilatation, and bilateral intra-abdominal testes. PBS has been rarely reported in sub-Saharan Africa.

The majority of pediatricians will rarely encounter a patient with PBS in their clinical practice. This article presents a case of a neonate seen in our institution (University Hospital of Kinshasa, Kinshasa, Democratic Republic of Congo) presenting with PBS.

2. Case report

A 4-day-old male neonate was referred to our center for follow up of a deficient abdominal wall musculature and fever.

Birth was through normal vaginal delivery with oligohydramnios. He was the second of two children. His brother was alive and in good health. There was no familial history of congenital disease. The mother was a 35-year-old woman, gravida 6, para 6. Her previous medical history and pregnancy course were uneventful. The mother reported no history of febrile illness, drug utilization, or ingesting herbal plants. Findings suggested there was no particular risk period or an environmental exposure before or during pregnancy. There was no history of consanguinity in the family. However, no antenatal ultrasound screening was performed during her pregnancy.

The physical examination at presentation showed a full-term neonate of 38 weeks’ gestation. The baby weighed 3.1 kg, with a height of 49 cm, and a cranial perimeter of 35 cm. There were no dysmorphic facies. The abdominal skin had an unusually wrinkled and flabby appearance resembling a dried prune (Figure 1). The perineum was smooth, without any anal opening. The baby had a normal phallus. Bilateral undescended testes were noted. Urine dribbling from a suprapubic vesico-cutaneous fistula below the urachus was observed.

Laboratory investigation revealed normal hematological parameters. Serum creatinine concentration was 415 μmol/L (26–88 μmol/L), serum urea was 35 mmol/L (2.2–11.3 mmol/L), serum sodium was 136 mmol/L (133–159 mmol/L), and serum potassium was 4.9 mmol/L (4.3–7.2 mmol/L).

Renal and abdominal ultrasonography revealed urinary tract anomalies characterized by bilateral hydronephrosis with dilated ureter and lack of visualization of the bladder.
with leakage of urine through the navel. Cardiovascular ultrasound was normal.

This clinical description is suggestive of PBS with persistence of the urachus. The child died from respiratory and renal failure 3 days after hospital admission.

3. Discussion

PBS is a rare congenital disease with an estimated incidence of 1/35,000–1/50,000 live births in Western countries. Approximately 97% of those affected by PBS are male.1

The diagnosis has been broadly inclusive by using clinical and morphologic features in combination. Clinical features of our patient were similar to those described in literature and were suggestive of PBS.1,4

The etiology of PBS is poorly understood. The deficient abdominal wall musculature associated with prune belly syndrome often results in numerous functional disabilities, including impaired bladder, urinary tract malformation, impaired bowel function, poor posture and balance, and in males, cryptorchidism. This embryological defect may have started between 6 and 7 weeks of gestation. The syndrome is caused by abdominal muscle deficiency secondary to a migrational defect of the lateral mesoblast or by urethral obstruction early in development, and it is the result of massive bladder distention and urinary ascites, leading to degeneration of the abdominal wall musculature and failure of testicular descent. The impaired elimination of urine from the bladder leads to oligohydramnios.1,4,5 Developmental delays and growth retardation have also been reported.

Prenatal diagnosis of PBS is based on ultrasound and is usually diagnosed in the second trimester. With prenatal ultrasonography, PBS can usually be diagnosed in the second trimester of pregnancy. In the present case, no ultrasonography was performed during the pregnancy.

In this context, PBS was diagnosed in the delivery room of a poor resourced settings.2,3 Postnatal ultrasonography is essential for the diagnosis of associated urinary tract anomalies.

There was no familial history of congenital disease in our case. The genetic cause of PBS was unknown. A possible relationship between PBS and HNF1β mutation was described in the literature.6,7 However, further genetic studies are necessary for the identification of the genetic basis of PBS. Facilities for genetic and molecular studies are not readily available in the Democratic Republic of Congo.

Treatment involves surgical correction of the abdominal wall defect and urinary tract abnormalities, early orchiopexy, and supportive management of associated defects.

In our case report, death occurred after 48 hours due to renal failure. Renal complications in a child with PBS are factors of high mortality.

4. Conclusion

Our case report serves to remind us of the difficulty of prenatal diagnosis and postnatal management of congenital diseases such as PBS in low-resource settings such as the Democratic Republic of Congo.

Conflicts of interest

The authors have no conflicts of interest relevant to this article.

References


