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# Prune-Belly Syndrome: A Case Report from Rwanda

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**Background: Prune-Belly syndrome, Eagle-Barret syndrome and triad syndrome, all refer to congenital anomalies involving abdominal musculature, urinary tract and testicles. The syndrome consists of a triad of abdominal muscle aplasia, massive ureteral and bladder dilatation and cryptorchidism. Kidneys are often affected by secondary hydronephrosis or by polycystic dysplasia. The full manifestation of the syndrome occurs almost exclusively in boys. Available reports on the epidemiology and outcome show a high perinatal mortality due to related prematurity and associated pulmonary complications. The management of a prune-belly patient has been controversial. However nowadays, the tendency is to assist primarily prune – belly neonates in respiratory failure, and to limit radical urologic interventions.**
**Case Report: We report hereby a full term prune-belly neonate who succumbed from this condition before being adequately investigated. Our purpose is to call clinicians' attention to early recognition, investigation and management of the syndrome. Even though infants with a full-blown syndrome have a poor prognosis for long term survival, all patients need careful evaluation and individualized management according to the spectrum of the syndrome. In this paper, the morphogenesis and the developmental biology of the abdominal wall will be also recalled, and literature reviewed.**

## Introduction

Prune – belly syndrome is characterized by absence, deficiency or hypoplasia of the abdominal musculature, accompanied by genitourinary abnormalities including a large hypotonic bladder, dilated and tortuous ureters, and bilateral cryptorchidism. In about 3 out of 4 patients with the prune belly syndrome, there are associated malformations of the cardiopulmonary, gastrointestinal, and orthopaedic systems[1]. The worldwide incidence ranges from 1 in 35,000 to 1 in 50,000 live births; but it is highly variable[2,3]. Because the condition is rare, there is scarcity of population-based epidemiology or mortality data available. Routh et al[4] in a recent epidemiologic nationwide study in the United States found an incidence of 3.8 / 100,000 live male births. The authors found a persistently high in-hospital mortality of 31% despite contemporary medical progress, and a high association of prematurity (43%) with consequent pulmonary co morbidity requiring ventilator support. Current management of a prune - belly baby is primarily supportive, with few surgical procedures[4,5]. Major surgical reconstruction tends to be reserved for severe urinary tract problems. Prognosis may vary from death in utero to a near-normal life expectancy[6].

## Case Presentation

A full term single pregnancy newborn male baby was referred to the Neonatology unit of Butare University Teaching Hospital, immediately after a vaginal delivery at a District Hospital. The neonate had a referral diagnosis of respiratory insufficiency and multiple congenital malformations. He was the first born of a young couple. On admission the baby was found not to be anaemic, jaundiced or cyanosed, but had poor saturation of 70% without oxygen supply. The baby had a pectus carinatum, and absence of antero-lateral abdominal muscles with a wrinkled skin. The peristalsis of the intestinal loops was visible through the skin (Figure 1).

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As a consequence of the aplasia of the rectus abdominis muscle, the anterior thoracic wall was tented, thus impairing the breathing movements hence the poor oxygen saturation (Figure 2). He also had a bilateral cryptorchidism. The micturition was normal. The neonate was treated with humidified oxygen supply at 4 litres/min by nasal prongs, and prophylactic antibiotics (Ampicilline 100 mg / Kg in three doses given every 8 hours intravenously, and Gentamycine 4 mg / Kg in two doses given every 12 hours intravenously). A surgical consultation was requested to have a clear diagnosis and the management plan. A diagnosis of Prune – belly syndrome was made and investigations had to begin with abdominal ultrasonography. Unfortunately the baby was discharged without the knowledge of the surgeon. The duty nurse reported that the mother had requested discharge in order to go and attend to her husband who 'had been involved in a road traffic accident' that occurred far away in the village. Whether that was a genuine reason (truth) or concocted story for demanding for discharge could not be established. The baby was reported to have died from his condition a few days later.

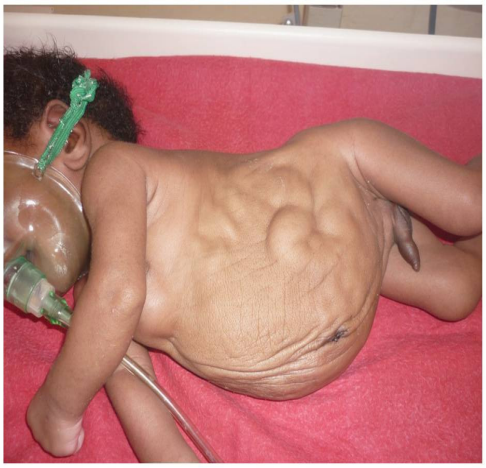

**Figure 1.** Visible intestinal loops through the skin.

**Figure 2,** Aplastic Rectus Abdominis Muscle,and Tented Anterior Thoracic Wall.

## Discussion

Although Prune –belly syndrome is a rare condition that clinicians should be able to diagnose. Prenatal diagnosis is possible sonographically as early as 13 to 14 weeks of gestation. Doppler imaging and 3-dimensional sonography were found to be a complementary method to conventional sonography[7,8] . To the best of our knowledge, no prenatal diagnosis of the syndrome has been reported in Rwanda or in the East African region. In neonatal period, paediatricians and neonatologists should make the diagnosis the condition, provide primary supportive treatment, and seek the urologist's advice.

The syndrome comprises genitourinary and extra genitourinary abnormalities. The central feature in the extra genitourinary abnormalities is the abdominal wall defect which affects characteristically the lower and medial parts. Myopathology reveals variation in muscle fibre diameter with haphazard arrangement of both atrophic and hypertrophic fibres, an increase in fibrous tissue surrounding the muscle fibres, and fatty infiltration. It is known that the abdominal wall is developed from the lateral mesoderm plate of the embryo which undergoes cavitation, splitting the plate into the somatopleure and the splanchopleure. Experimental studies show that the BMP-4 is the signalling molecule for the lateral mesoderm plate[9]. It has been recently shown that mutation of FGFr1 and FGFr2 mutation in embryo disrupt dermal and muscle development of the anterior abdominal wall[10]. The FGF is also involved in development of urogenital system[11]. Thus, although the cause of Prune Belly Syndrome is still unknown, it is clear that common developmental factor for the anterior abdominal wall and urogenital system is involved. The poor support of the lower chest wall by a wrinkled abdominal wall impairs effective cough mechanisms,

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rending patients vulnerable to respiratory infections. Other extra genitourinary abnormalities vary widely from a region to another. While Routh et al[4] found these to be mainly pulmonary (55%), gastrointestinal (31%) and cardiac (10%) in America, Salihu et al[12] in Cameroon identified clubfoot (45%), pulmonary hypoplasia (27%), Potter facies (27%), imperforate anus (27%), and arthrogryposis (18%), among 11 cases over a 13 year period.
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Genitourinary abnormalities may affect the urinary tract from kidneys to the urethra. Renal dysplasia and hydronephrosis are the most common renal abnormalities. Dysplasia may account for 50% of cases. The greatest threat to renal parenchyma is recurrent infection. Ureters are tortuous with segments of massive dilatation, the distal part being the most affected. The bladder is grossly distended with a large urachal diverticulum which, if patent, can be used as an alternative to a cystostomy to divert urine, providing with better cosmesis and quality of life[13]. Following these distension and enlargement there is a consequent vesicoureteral reflux in up to 85%, and reduced detrusor contractility. The bladder neck is wide, the prostate hypoplasic, and the prostatic urethra grossly dilated; hence the dilated radiographic appearance. A common pathophysiological feature of the urinary tract is a marked decrease in epithelial cells, decrease in smooth muscle fibers and increase in connective tissue. Bilateral cryptorchidism is a central anomaly in the genitourinary abnormalities. In most cases testes are intraabdominal. Obstructed descent is thought to be due to the dilated urinary system and an intrinsic abnormality in the testes.

Prune-belly patients can be divided into three categories correlating with the outcome. Category one neonates have a severe pulmonary or renal dysplasia that precludes survival beyond the first days of life. The most affected are stillborn. Those with the potential to survive the neonatal period classify in category two. They have typical external features and uropathy of the full-blown syndrome but no immediate problem with survival. Despite lack of appropriate work-up, we think the reported patient to belong to this class. Category three includes those with mild or incomplete features of the syndrome. For these, renal function is stable, uropathy less severe. They may present in adulthood[14].

The management of prune-belly neonates consists primarily of stabilization of the cardiorespiratory function, and then urological evaluation proceeds. Surgical correction of urinary stasis and reflux is performed after pulmonary maturation. This entails: ureteral shortening, vesicoureteral reimplantation and reduction cystectomy performed in conjunction with abdominal reconstruction. The conservative approach to the management of urinary tract dilatation can be limited to nephrostomy or nephroureterostomy[5].

Proponents of limited surgical intervention advocate surveillance, management of infection, and surgery only in evident obstruction, high grade reflux or intractable infection. Orchidopexy is performed during infancy, but the potential for fertility is compromised.

## Conclusion

Prune-belly syndrome is a disease with a broad spectrum of presentation. The prognosis depends on the degree of prematurity, pulmonary complications and renal dysplasia. The management of prune-belly neonates is primarily stabilization of the cardio respiratory function. Urine diversion is preferred to extended urological reconstruction. Abdominal wall reconstruction gives good cosmetic results to patients and psychological effect to parents. These children require long-term follow-up. Up to now, the precise form of any genetic inheritance for PBS is still unknown. It may be variable and it may often represent a new mutation. A sex linked autosomal recessive form of inheritance and other genetic basis has been strongly suggested[15,16].

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