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CASE REPORT

Prune Belly syndrome with vesicocutaneous fistula: a rare case report and literature review

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Abstract

Prune Belly syndrome (PBS) is a rare complex malformation that preferentially affects male newborns. It presents with a wide spectrum of alterations that include abdominal muscle deficiency, varying degrees of renal dysplasia and urinary tract obstruction, in addition to bilateral cryptorchidism. In general, it is associated with other developmental anomalies, mainly of cardiac, pulmonary and gastrointestinal origin. Despite numerous advances in the treatment of complex malformations, morbidity and mortality in PBS remains high, due to early renal failure and associated malformations. This is a case report of a male newborn, inserted in a complex social context, born by vaginal delivery, premature, with a gestational age of 36 weeks and 1 day. At the initial assessment in the delivery room, he presented: male external genitalia, bilateral cryptorchidism, flabby and hollowed abdomen, parchment skin suggestive of the absence of abdominal muscles, mild respiratory effort and bradycardia. He received positive pressure ventilation and was referred to the Neonatal Intensive Care Unit (UTIn) where a vesicocutaneous fistula and renal failure were found, despite the vesicostomy, progressing to death at 1 month and 1 day of chronological age. Complex congenital malformations, such as PBS, require individualized assessment so that better postnatal results are achieved. However, the limitation of therapeutic care must be carefully evaluated, considering the great morbidity that accompanies these patients. This article aims to describe a rare case of PBS, detailing the evolution of patients with this malformation.

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INTRODUCTION

Prune belly syndrome (PBS) is a rare congenital disease that affects approximately 3.8 per 100,000 live births.[1,2] It is more prevalent among male newborns, who account for approximately 95% of affected patients. It is characterized by the clinical triad of deficient abdominal musculature, genitourinary tract abnormalities and bilateral cryptorchidism. Urinary tract changes vary substantially in severity and include renal hypoplasia or dysplasia, hydronephrosis, ureteral ectasia, bladder dilation and urethral obstruction. On ectoscopy examination, the abdomen is often described as having a prune-like appearance due to the partial or total absence of the abdominal wall muscles.[1.2]

More than 50% of the subjects with PBS die and most develop comorbid conditions, with 67% showing kidney failure and 48% requiring mechanical ventilation in neonatal life. Morbidity and mortality are further aggravated by pulmonary, skeletal, cardiac and gastrointestinal defects, present in approximately 75% of the cases.[2,3,4]

The etiology of the syndrome is not fully understood, but concordance between monozygotic twins (12.2/100,000 live births) points to a probable genetic association. About 3% of the patients diagnosed with PBS present hepatocyte nuclear factor (HNF1β) mutations, which is involved in the differentiation of the mesoderm and ectoderm in embryonic development.[5] According to this theory, anomalies arise from the abnormal development of the first lumbar myotome, between 6 and 10 weeks of gestation, resulting in hypoplasia or agenesis of the abdominal muscles, in addition to urinary tract abnormalities. Another hypothesis suggests that malformations and obstructions of the urethra trigger distension of the pyelocaliceal system, preventing adequate development. Bladder distension might further interfere with testicular descent, causing bilateral cryptorchidism. The higher incidence of the syndrome in males and the variety of urinary tract alterations might be explained by the more complex morphogenesis of the male urethra.[6]

This article reports a case of PBS and describes the numerous signs and symptoms and particularities of clinical management through a review of current literature.

CASE REPORT

A G3P2A0 woman, pregnant for 36 weeks and 1 day according to obstetric ultrasound examination performed in the 2nd trimester, was hospitalized in preterm labor. She admitted to using cocaine, marijuana, risperidone, clonazepam and fluoxetine during pregnancy. She did not undergo proper prenatal care and presented a prenatal care card with negative serology for HIV, syphilis, hepatitis B, toxoplasmosis and rubella in the 2nd trimester of pregnancy and an obstetric ultrasound report showing polyhydramnios, a fetus with massive ascites, thoraco-abdominal disproportion, bilateral pyelocaliceal dilation and a non-visualized bladder. She added that she had two healthy children.

Upon physical examination, the woman in labor had signs of uterine dynamics and a cervix dilated to 8 centimeters, which developed into a spontaneous vaginal birth without complications. Assessment of the newborn indicated the presence of male external genitalia, bilateral cryptorchidism, a flaccid and hollow abdomen with parchment-like skin, suggestive of absence of abdominal muscles, mild respiratory effort, and bradycardia. The patient responded well to the introduction of positive pressure ventilation. (Figure 1)

The newborn was taken to the ICU where he was placed on invasive mechanical ventilation under continuous sedation. Laboratory tests on admission showed normocytic normochromic anemia, thrombocytopenia, hyperkalemia (5.0 mEq/L), elevated urea (22 mg/dL) and creatinine (1.5 mg/dL). On the second day of life, he presented with anuria, drainage of urine through the umbilical stump and deterioration of kidney function, with worsening of hyperkalemia (6.0 mEq/L), urea (26 mg/dL) and creatinine (2.8 mg/dL).

Ultrasound scans of the urinary tract revealed dilation of the pyelocaliceal system on the left. The bladder was not fully visualized.



Figure 1. Prune belly

During hospitalization, the newborn developed urinary tract infection, hemorrhagic complications, hemodynamic instability refractory to vasoactive drugs, progressive worsening of kidney function with respective urea and creatinine levels of 167 mg/dL and 3.48 mg/dL, which caused the postponement of a previously scheduled vesicostomy. The patient eventually underwent the procedure, but his kidney function did not improve after it.

An echocardiogram performed to search for associated malformations found an ostium secundum atrial septal defect of moderate size, a moderate patent ductus arteriosus and moderate enlargement of the right atrium and ventricle.

Since his kidney function worsened and he needed renal replacement therapy, a multidisciplinary team evaluated the newborn for a potential transfer to a referral hospital. However, he remained unresponsive to neurological examination without sedation and could not be weaned from mechanical ventilation. In view of the patient's poor prognosis, the decision was made, with the involvement of family members, to limit the therapeutic efforts aimed at the patient. He died one month and a day after birth.

DISCUSSION

PBS is characterized by a broad spectrum of signs and symptoms. Woodard divided these manifestations into three major categories of neonatal presentations. Category I, which is rarer, results from severe renal hypoplasia or severe urethral obstruction and includes severe oligohydramnios, pulmonary hypoplasia and Potter's facies. Category II includes the complete clinical triad and generally progresses to kidney failure due to moderate renal dysplasia. Category III is characterized by mild clinical disease, without renal dysplasia or pulmonary hypoplasia.[5,7]

Diagnosis is often performed after birth, although obstetric ultrasound examination, especially when performed by a trained professional, might detect changes suggestive of PBS in the 10th to the 13th week of pregnancy, thus allowing early therapeutic planning in the neonatal period.[1,8,9] Therefore, fetal ultrasound is even more important in pregnant women at higher risk for malformations, such as consanguineous couples and individuals with a family history of urogenital anomalies.[4,5]

Ultrasound examination may also indicate the presence of factors that directly influence the prognosis of patients, such as the presence of renal dysplasia, urinary tract obstruction, oligohydramnios and associated malformations.[10] Pulmonary hypoplasia must also be considered, since from the 8th to 10th week of gestation the kidneys begin to excrete urine to form the amniotic fluid, which is essential for fetal lung development.[9,11]

In selected cases, intrauterine treatment of complications including urinary drainage procedures is possible. Vesicoamniotic shunting decreases the dilation of urinary tract structures, contributes to improved structural development of the kidneys and prevents pulmonary hypoplasia.[1,9]

Despite the scarcity of data in medical literature, this article draws attention to a possible association between PBS and use of cocaine on account of the intense vasoconstriction of the uteroplacental vessels induced by the drug, which may compromise the differentiation of the mesoderm and ectoderm in the first weeks of embryonic development.[12,13]

This article describes the case of a newborn with category II PBS. Polyhydramnios – a key factor in fetal lung development – in the second trimester ultrasound scans is an important finding attributable to the presence of a vesicocutaneous fistula.[11] The mother did not undergo proper prenatal care during pregnancy due to sociocultural complications, which made it impossible for the attending medical team to develop a treatment plan.

Postnatal treatment of PBS aims to preserve kidney function. About 75% of the patients have an enlarged, hypotonic bladder, which can store urine but lacks full emptying ability. Therefore, early vesicostomy is essential to prevent the deterioration of kidney function. Urinary retention is also an important factor in recurrent urinary tract infections. Prophylactic antibiotic therapy is usually recommended to reduce additional damage to the kidneys. However, despite therapeutic efforts, 40–50% of the patients develop kidney failure and require renal replacement therapy.[3,5]

Although some patients are eligible for urinary tract and abdominal wall reconstruction surgery, potential pulmonary and cardiac alterations associated with PBS may cause respiratory failure and sudden death after general anesthesia, making surgery a less viable option for individuals with the condition.[14]

As described in this case report, the high morbidity and mortality associated with PBS often impose the need to limit therapeutic efforts. Palliative care, especially in pediatric ICUs, encounters cultural barriers in the difficulty of dealing with death.[15] For families, the loss of a child tends to be more devastating than the death of an adult. For care teams, prescribing palliative care is often synonyms with failure.[15] In neonatology, critically examining the treatments prescribed to patients with rare diseases and complex clinical conditions is more important than in other areas of medicine, since despite the increases in survival achieved via advances in care, the comorbid conditions these patients develop are significant and, at times, limiting.

CONCLUSION

Like other congenital syndromes, PBS requires early diagnosis and an individualized approach to achieve better outcomes. This article reported a case with an unfavorable outcome due to progression to renal failure and drew attention to the importance of prenatal diagnosis and palliative care in pediatrics.

REFERENCES

1. Achour R, Bennour W, Ksibi I, Cheour M, Hamila T, Hmid RB, et al. Prune belly syndrome: Approaches to its diagnosis and management. Intractable Rare Dis Res. 2018;7(4):271–4.
2. Kheir AEM, Ali EMA, Medani SA, Maaty HS. Prune belly syndrome: A report of 15 cases from Sudan. Sudan J Paediatr. 2017;17(1):42–8.
3. Carvalho NDS, Cavachini C, Dudus MM. Síndrome de Prune Belly. Resid Pediatr. 2018;8(1):45-7
4. Granberg CF, Harrison SM, Dajusta D, Zhang S, Hajarnis S, Igarashi P, et al. Genetic Basis of Prune Belly Syndrome: Screening for HNF1β Gene. J Urol. 2012;187(1):272–8.
5. Arlen AM, Nawaf C, Kirsch AJ. Prune Belly syndrome: current perspectives. Pediatric Health Med Ther. 2019;10:75–81.
6. Tagore KR, Ramineni AKS, Lakshmi ARV, Bhavani N. Prune Belly Syndrome. Case Rep Pediatr. 2011;2011:1-3.
7. Samal SK, Rathod S. Prune Belly syndrome: A rare case report. J Nat Sci Biol Med. 2015 Jan-Jun;6(1):255–7
8. Alkhamis WH, Abdulghani SH, Altaki A. Challenging diagnosis of prune belly syndrome antenatally: a case report. J Med Case Rep. 2019 Jun;13:198.
9. Byon M, Kim GJ. Prune-belly syndrome detected by ultrasound in the first trimester and the usefulness of vesicocentesis as a modality of treatment. Obstet Gynecol Sci. 2013;56(4):265–8.
10. Brown PSH, Hines RS, James R. Prune belly syndrome in adult man. Uro. 1993 Feb;41(2):199.
11. Sarhan OM, Al-Ghanbar MS, Nakshabandi ZM. Prune belly syndrome with urethral hypoplasia and vesico-cutaneous fistula: A case report and review of literature. Urol Ann. 2013 Oct-Dec;5(4):296–8.
12. Mahalik MP, Gautieri RF, Mann Jr DE. Teratogenic potential of cocaine hydrochloride in CF-mice. J Pharm Sci 1980;69(6):703-6
13. Greenfield SP, Rutigliano E, Steinnhardt G, Elder JS Genitourinary tract malformations and maternal cocaine abuse. URO 1991;37(5):455-9.
14. Grimsby GM, Harrison SM, Granderg CF, Bernstein I, Baker LA. Impact and frequency of extra-genitourinary manifestations of prune belly syndrome. J Pediatr Urol. 2015 Oct; 11(5): 280e1–6.
15. Marçola L, Barbosa SMM, Zoboli I, Polastrini RTV Ceccon MEJ. Analysis of death and palliative care in a neonatal intensive care unit. Rev. Paul. Pediatr. 2017 Apr-Jun;35(2):125-9.