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ORIGINAL ARTICLE

Pseudo Prune Belly syndrome in female patient: a case report

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Abstract

Pseudo Prune Belly syndrome (PPBS) corresponds to an incomplete form of Prune Belly syndrome (PBS), which does not fill the triad composed of abdominal wall muscle alterations, bilateral cryptorchidism and abnormalities of the urinary tract. Patients with partial abdominal alterations, absent or unilateral cryptorchidism, and female are classified as having PPBS. PBS was a rare condition, with an incidence of 1/40,000 live births, and it is even more uncommon to find patients with PPSB, as it affects 3-5% of cases initially diagnosed with PBS. The following case reports a 1-year and 9-month-old female patient, allegedly with PBS from birth, admitted because of an infectious cause, and who, during hospitalization and complementary evaluation, had her diagnosis changed to PSPB. Due to underdiagnosis and misdiagnosis of the PPBS, the report aims to promote acknowledgement about this disease, reinforce the clinical diagnosis from the most prevalent findings and ratify the importance of early therapy as a determinant of quality of life. In addition, the study brings to discussion the most involved prognostic factors and possible complications related to both late diagnosis and inadequate management of these patients.

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INTRODUCTION

Prune syndrome Belly , also called Eagle Barrett Syndrome[1] or "dry plum belly" syndrome, is clinically diagnosed at birth, by the triad: absence or hypoplasia of abdominal muscles, cryptorchidism and urogenital changes. The term "dry plum belly" is a popular name for the syndrome and refers to the wrinkled appearance of the abdomen caused by changes in the muscular wall. Patients with partial abdominal alterations, absent or unilateral cryptorchidism and female patients are classified as having Pseudo Prune Syndrome. Belly (PSPB). The cases described in female patients are even rarer[1].

The most described findings are related to urogenital changes, especially hydronephrosis (75% of patients), renal dysplasia, hydroureter , renal cysts and vesicoureteral reflux[3]. In about 23-25% of cases, there is an association of involvement of other systems, especially gastrointestinal, skeletal (geno varus, pectus excavatum), cardiac and pulmonary[1-3]. Significant abdominal involvement may be the cause of cardiorespiratory changes[1]. A common and different finding is the high implantation of the umbilicus[1,2].

Regarding the diagnosis, some studies suggest that it should be clinical, and the radiologist's role is to assess the extent of organic involvement to determine surgical interventions, and the pathologist's role to assess the severity of renal dysplasia, one of the main prognostic factors[1,2].

The treatment and follow-up of these patients must be individualized, always aiming at maintaining quality of life and reducing the risk of progression to chronic kidney disease[1].

CASE REPORT

YVG, 1 year and 9 months old, was admitted to the emergency department for sepsis of a pulmonary focus. After clinical evaluation, the patient was in a serious general condition, with wrinkled skin on the abdomen, bilateral prominence of costal margins, ambiguous genitalia, imperforate anus, severe kyphoscoliosis, laparotomy scar, vesicostomy and colostomy ostium (Figures 1 and 2).

According to information collected, the patient was diagnosed with PBS at birth, underwent vesicostomy surgery and an exploratory laparotomy, due to genitourinary and anorectal abnormalities (complex cloaca, persistent urachus) inherent to the syndrome, as well as to colostomy, a gastrointestinal characteristic related. She also has significant spinal deviation and congenital clubfoot, skeletal disorders related to the condition, for which the conduct is conservative at this age. Ambiguous genitalia is not closely related to the syndrome, but would define the diagnosis, SPB or PSPB. In order to clarify the sex, diagnosis and appropriate therapy, karyotype, ultrasound of the abdomen (US) (not conclusive since the testicles, uterus and ovaries), magnetic resonance imaging (MRI) of the abdomen and urinary tract and angiography were requested.



Figure 1. Imperforate anus and ambiguous genitalia that led to doubt about the patient's sex.

computed tomography (AngioCT) of the chest. The karyotype result was 46XX, characterizing female sex. The MRI showed changes related to the underlying disease, highlighting: bone and abdominal wall malformations. In the pelvis, the path of the urethra, vaginal canal and rectal ampulla were not identified, suggesting the presence of a cloaca. A thickened bladder wall and bilateral kidneys were also visualized with several alterations to be investigated by other methods. There were no testes, but there is a cystic image in the topography of the bladder and a tissue in the pelvic floor on the right, laterally to the kidney, of undetermined origin. CT angiography showed no noteworthy changes. To date, investigations of the urogenital alterations had not been completed, but as the patient was in conditions of medical discharge, outpatient follow-up was recommended. Thus, from the karyotype result, it was concluded that the patient's sex is female, corroborating the diagnosis of PSPB, a rare entity in this gender. Regarding the pulmonary condition, which is why she was hospitalized, she developed septic shock, requiring the use of vasoactive drugs and orotracheal intubation. She had negative blood cultures throughout her hospital stay and positive urine culture for Pseudomonas, Candida and Proteus, successively, all of which were treated with appropriate antibiotic therapy guided by an antibiogram. She evolved with resolution of the septic condition, with pulmonary improvement limited by the anatomy and thoracic mechanics, and prophylaxis for urinary tract infection was started with trimethoprim-sulfamethoxazole. She was discharged with referrals to outpatient pediatrics, orthopedics, nephrology, pulmonology, gastropediatrics, and pediatric surgery.



Figure 2. Abdominal changes ("plum belly"), colostomy ostium and laparotomy scar hiding vesicostomy.

DISCUSSION

A literature review was carried out in the main databases, with the terms " Pseudo prune belly Syndrome ", and only 7 articles were found in Pubmed and 2 articles in Science Direct (one of them also found in Pubmed), among case reports and literature reviews. The data obtained were related to the reported clinical case.

PSPB is a rare entity that leads to several functional alterations in affected patients. The incidence of SPB is around 1/40,000 live births[1]. However, it is estimated that 3 to 5% of patients with SPB actually have PSPB, despite the few case reports available, and it is believed to be a phenotypic variant of SPB[1,3].

There are two theories that aim to explain the pathophysiology of SPB and its variant. The oldest of them postulates that the changes are due to prenatal obstructive processes leading to urinary tract dilation and abdominal distension in the fetus. The process then culminates in abdominal hypoplasia. However, this theory has been replaced as it does not explain the large amount of extra-abdominal and extra- urinary changes associated with the syndrome. Thus, the theory of lateral mesoderm migration defect emerged, between 6 and 10 weeks of gestation, which would explain the urological, testicular and abdominal alterations[1,2].

In the case described, the patient was admitted to the service with a diagnosis of PBS and in the presence of infection of a pulmonary and urinary focus. The infectious condition was treated during hospitalization. The karyotype confirmed the female sex, concluding the diagnosis of PSPB. From this, the investigation of the changes that the patient had to institute therapy with the objective of providing a better quality of life was initiated.

Regarding treatment, 38% of patients need ureteral reimplantation and 25% need an ileal canal[3]. There are no data on the percentage of cutaneous vesicostomies and sphincterotomy , but they are tools described as alternatives to optimize the drainage of the urogenital system and try to preserve renal function for a longer time[2]. Despite all attempts, 63% of patients with PSPB will progress to chronic kidney disease (CKD)[2]. The main factors associated with progression to CKD are renal changes on ultrasound, creatinine above 0.7 mg/ dL and recurrent pyelonephritis[4]. The other interventions will be evaluated according to the need presented by PSPB patients, however, the abdominal wall reconstruction offers functional benefits such as improved lung, heart and bowel function, in addition to aesthetic benefits[5]. In the case described, the companion mentioned that she had undergone vesicostomy and laparoscopy for hysterosalpingectomy at birth, without a summary of discharge for confirmation. After admission to the Pediatric Care Unit for the treatment of sepsis with a pulmonary focus, he underwent only imaging tests to analyze the malformations and define the next clinical and surgical procedures. The patient is under outpatient follow-up at the same service.

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