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Prune Belly Syndrome: A Case Report

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Abstract

Background: Prune belly syndrome is a rare congenital disorder characterized by a triad of deficient abdominal wall muscles, urinary tract anomalies and cryptorchidism. Most of the affected cases are males. This syndrome is associated with a high mortality rate in the peri-natal period due to pulmonary complications. We present the first prune belly syndrome case in Lebanon. Besides, despite the presence of bilateral urinary tract dilation, no pulmonary consequences emerged at birth.

Case Presentation: A case of a term baby boy born to a 30 years old woman was found to have floppy abdominal wall and mild tachypnea so was admitted to the neonatal intensive care unit for investigations and management. Physical examination revealed bilateral undescended testicles. Imaging reported bilateral severe hydronephrosis with ureteral dilation and absence of ventral abdominal musculature. So, a diagnosis of Prune belly syndrome was established. Due to evidence of kidney injury, supra-pubic cystostomy was performed to relieve the obstruction. This lead to improvement in kidney function test. Patient was planned for later repair of abdominal wall and for orchiopexy.

Conclusion: The case was a very interesting experience for the whole team especially due to the absence of clear guidelines due to the rarity of the syndrome. It highlights the importance of shared decision between the neonatologists and the urologists in any unusual disorder. Moreover, it shows the significance of pre-natal imaging and the necessity of searching for associated anomalies in the case of severe hydronephrosis pre-natally.

Keywords: Prune belly syndrome; Abdominal musculature; Bilateral hydronephrosis; Newborn

Introduction

Prune belly syndrome (PBS) which is also known as Eagle Barrett Syndrome is a congenital disorder characterized by the presence of a classic triad: flaccid hypoplastic skeletal muscles of the ventral abdominal wall resulting in prune-like structure, megacystis with megaureter and poor smooth muscles contraction, and bilateral intra-abdominal cryptorchidism. This extremely rare congenital myopathy affects about 1 in every 25,000 individuals [1]. Prune belly syndrome affects boys primarily with females representing less than 5% of total cases [2]. It constitutes a disease spectrum with severity classified according to the degree of renal dysplasia and pulmonary compromise.

There is no clear cause behind PBS but the presence of the disease in monozygotic twins, in families and the higher incidence in males suggests a genetic underlying factor to be further studied.

Two theories were proposed to explain the malformation. The first one explains a defect during mesenchymal formation resulting in maldevelopment of abdominal wall muscles and urinary tract. The other theory suggests an in-utero urinary tract obstruction at the level of urethra or prostate resulting in dilation of urinary tract upstream the obstruction ending by secondary maldevelopment of abdominal wall muscles [2].

Due to the rarity of the syndrome, no clear guidelines are present for the diagnosis and management. Although some cases reported full pre-natal diagnosis by pre-natal sonography, not all cases can be diagnosed prenatally although presence of pre-natal sonographic findings suggesting lower urinary tract obstruction may point toward such diagnosis.

Below we describe a newborn male with the extremely rare prune belly syndrome.

Case Presentation

A case of term baby boy born by caesarean section to a 30 year old woman. APGAR score at birth was 9 and 10 at 1 and 5 minutes after birth respectively. Soon after birth he started having mild subcostal retractions so was admitted to the neonatal intensive care unit for observation and investigations of the floppy abdominal wall shown in Figure 1.

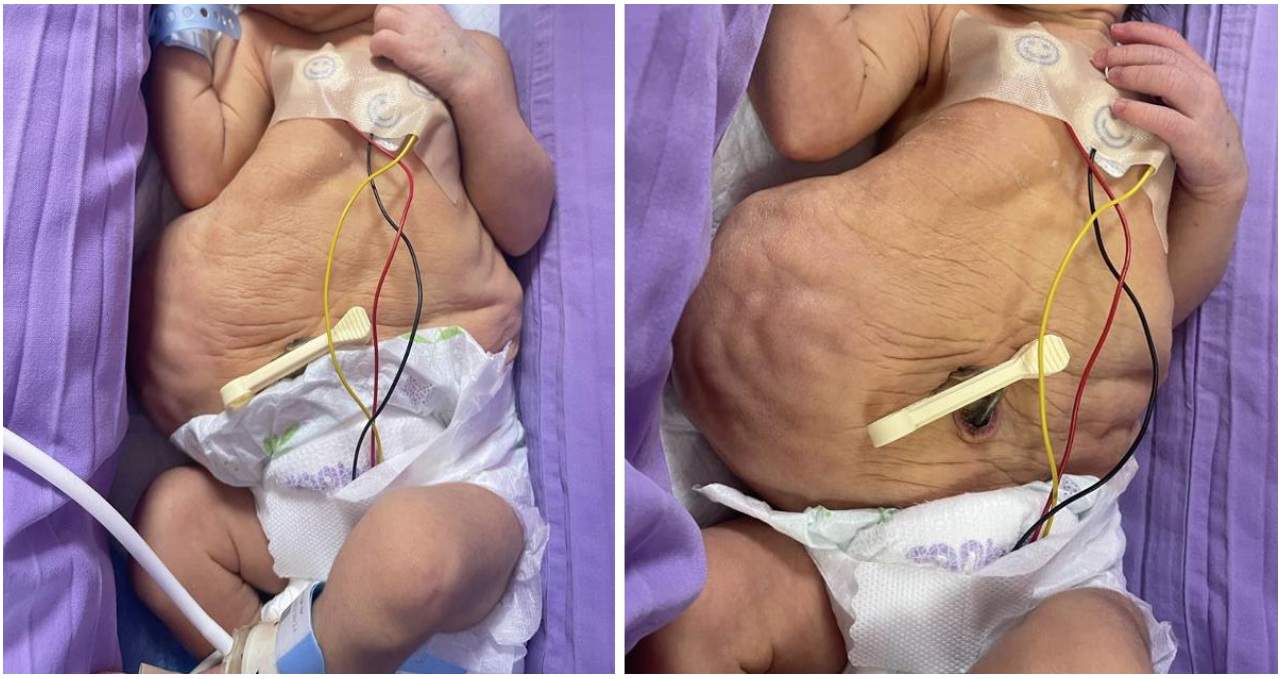

Figure 1: Abdominal wall of the newborn (photo taken soon after birth).

Maternal history reveals a 30 years old previously healthy woman, G3P3A0. Her pregnancy was followed by a gynecologist and showed no history of infections or gestational complications. A prenatal ultrasound performed suggested bilateral hydronephrosis with abdominal mass that give the impression of abdominal duplication cyst or ascites. Post-natal follow up was advised at that time. Physical examination revealed a 2.9 kg baby boy with non-palpable testes bilaterally and deficient abdominal musculature.

Systemic review of the baby showed a transient tachypnea of newborn that resolved within hours after requiring 0.5 l/min oxygen by nasal canula. A Chest X-ray was performed and revealed normal pulmonary parenchyma and cardiac silhouette with mild peri-hilar congestion. A trans-thoracic echocardiography done at day of life (DOL) 5 was normal.

As soon as the respiratory condition improved, the baby was started on oral term formula which was tolerated. The first meconium passed within the first 24 hours after birth. Abdomino-pelvic ultrasound was performed as part of the post-natal follow up. It showed severe bilateral hydronephrosis. There was a normal hepatobiliary system and spleen. Cystic intestinal dilatations were detected just underlying the cutaneous layer with absent abdominal wall muscles along the bilateral bulging flanks.

This was followed on the next day by computed tomography scanner (CT-scan) abdomen-pelvis with intravenous contrast that confirmed the presence of severe bilateral hydronephrosis and severe dilatation of both ureters accompanied by severe circumferential urinary bladder wall thickening. Besides, absence of anterior abdominal wall musculature was detected which highly suggested the diagnosis of prune belly syndrome.

Based on the imaging done above, specialists consultation was recommended. Regarding the abdominal wall musculature, the pediatric surgeon suggested repair at age of 6 months. On the other hand, the function of the kidney showed persistent hematuria since birth with progressive deterioration during the first week of life with an increase in serum creatinine from 1.16 mg/dL at day one of life to 1.3 mg/dL at day 4 of life. The consulted urologist recommended voiding cystourethrography (VCUG) which showed narrowing of the posterior urethra and reflux at the level of the hypogastric loops.

At day of life 6 (DOL 6), a vesicostomy (suprapubic cystostomy) was performed under general anesthesia. The patient recovered successfully with a serum creatinine value of 0.6 mg/dL at day 3 post operation. He was discharged at DOL 11 after completing 5 days of post-operative prophylactic antibiotics and planned for abdominal wall repair by age of 6 months and orchiopexy with circumcision by age of 10 months.

Discussion and Conclusion

Prune belly syndrome was first described by Frolich in 1839. It acquired its name due to the wrinkled appearance of the abdomen [3]. The mortality rate in live newborns with PBS is estimated to be 20% [3]. Woodard described three categories for classification of neonatal presentation of PBS [4]. Type I is described in a patient with severe oligohydramnios pre-natally due to the significant renal dysplasia which will lead to pulmonary hypoplasia and potter facies and this will eventually end up by neonatal demise soon after birth.

The second type to which our described patient belongs presents with full triad including moderate renal dysplasia but no pulmonary hypoplasia. The third type is a mild form with no renal dysplasia, no pulmonary hypoplasia and normal renal function [4]. Early urologic intervention must focus on the relief of the urologic obstruction to preserve the kidney function which was done in the case described above by early vesicostomy. Circumcision and prophylactic antibiotics are also two suggested management steps to preserve kidney function by avoiding recurrent urinary tract infections [2].

The case described highlights the importance of multi-disciplinary approach to complex neonatologic cases. Moreover, the significance of early intervention on improvement of kidney function tests. And lastly, it shows the importance of search for associated anomalies when faced with a case of severe bilateral hydronephrosis pre-natally.

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