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Prune Belly Syndrome: A Case Report from Barranquilla, Colombia

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Abstract: ***Prune Belly syndrome, also known as Eagle-Barrett syndrome, is a rare congenital malformation characterized by hypoplasia of the abdominal musculature, cryptorchidism, and abnormalities in the urinary tract. Treatment is generally conservative, with surgical interventions considered in specific cases based on the severity of organ involvement. This report presents the case of a 37-week-old male newborn diagnosed with Prune Belly syndrome, confirmed through clinical evaluation and ultrasound. This case emphasizes the critical role of early diagnosis and multidisciplinary management in improving patient outcomes and quality of life.***

Keywords: Prune Belly syndrome, cryptorchidism, abdominal wall, urinary tract

1. Introduction

Eagle-Barrett syndrome, also known as Prune Belly syndrome (PBS), is a rare congenital malformation of uncertain etiology, primarily characterized by a clinical triad: hypoplasia or absence of abdominal musculature, bilateral cryptorchidism, and urinary tract abnormalities [1]. This condition, giving the abdomen a wrinkled, loose appearance similar to a “prune,” predominantly affects males, with an estimated global incidence of 1 in 30,000 live births. In Latin America, the incidence is approximately 3.8 cases per 100,000 live male births, making it relatively uncommon. [2]. Early recognition and treatment are vital to prevent significant morbidity and mortality associated with this syndrome [7]. A male-linked inheritance pattern is suspected, although familial cases are rarely reported. Skeletal abnormalities associated with oligohydramnios may occur, but lower limb abnormalities are rare. [4]

The triad characterizing this syndrome was first described by Parker in 1895, linking it to urinary tract malformations. However, in 1839, Frolich was the first to describe this syndrome in a child with abdominal musculature deficiency, keel chest, and absence of testicular descent. Subsequently, in 1901, William Osler described it as “Prune Belly Syndrome” due to the abdomen’s lax, wrinkled appearance. In 1950, Eagle and Barrett reported nine cases involving abdominal muscle absence associated with genitourinary abnormalities.

Prune Belly syndrome is infrequent and includes undescended bilateral testicles and urinary tract abnormalities. Although patients with this syndrome typically have normal or enlarged bladders, they may experience poor emptying. Ureters tend to be dilated and may have peristaltic abnormalities, and the urethra may be narrow or stenotic in some patients. The management of this syndrome has improved due to a better understanding of its pathophysiology; however, it remains challenging, especially in low-resource settings. This report presents a case of a 37-week-old male newborn identified with Prune Belly syndrome in a neonatal intensive care unit in Barranquilla, Colombia.

2. Methodology

T The methodology involved a comprehensive review of the patient's medical history and relevant findings from databases such as PubMed, Medline, and Embase. This review provided context for the case and helped establish connections between clinical presentation and established literature.

3. Case Report

A 37-week-old male newborn (Ballard score), the son of a 23-year-old primigravida mother with blood type O positive, no consanguinity with her partner, and a history of hypertensive disorder of pregnancy, gestational diabetes with unknown metabolic control, and trichomonas vaginitis. Obstetric ultrasound reported a deformed abdominal circumference potentially indicating a megabladder; however, despite these prenatal diagnoses, there was no documented follow-up by the gynecology team. The patient was born at a second-level hospital in Barranquilla via vaginal delivery, with Apgar scores of 8/10 at one minute and 9/10 at five minutes. His anthropometric measurements were appropriate for his gestational age: weight, 2,680 grams; length, 48 cm. On physical examination, the newborn exhibited dysmorphic facies: low-set ears, abdominal wall muscle deficiency, and bilateral cryptorchidism. Based on ultrasound findings, reports of in-utero urinary tract malformations, and physical exam details, a PBS diagnosis was made.

Upon admission to the neonatal intensive care unit at the Adelita de Char Mired IPS Hospital, the patient was in fair general condition, hemodynamically stable, but with marked abdominal distension. A CT scan of the abdomen and urinary ultrasound revealed absent abdominal rectus muscles, a distended bladder with thin walls, and grade IV bilateral hydronephrosis with megaureters. A general surgery

consultation recommended conservative management until nephrology could determine the course of action. Nephrology suggested placing a permanent urinary catheter due to renal injury secondary to obstructive uropathy. Follow-up ultrasound after 72 hours showed a decreased left kidney size with mild bilateral hydronephrosis and declining renal function, leading to catheter removal with retained and adequate urine output. Pediatric urology consulted and requested a urotomography showing bilateral hydronephrosis. No surgical indications were determined, and the patient was discharged on the 14th day of life with follow-up in previously mentioned specialties and exam monitoring.

Figure 1: Patient with Prune Belly syndrome, absent abdominal wall muscles, distended abdomen, thin wrinkled skin, bilateral cryptorchidism, low-set ears.

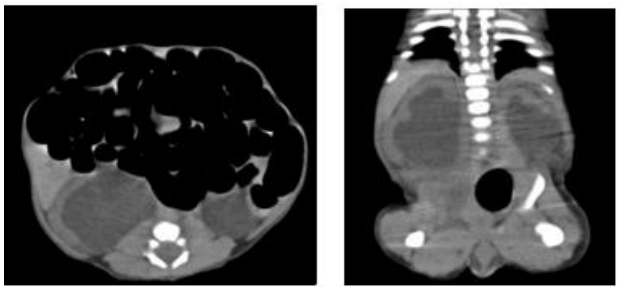

Figure 2: Urotomography showing bilateral hydronephrosis

4. Discussion

Prune Belly syndrome (PBS), also known as Eagle-Barrett syndrome, is a congenital disorder characterized by a clinical triad of abdominal muscle deficiency, severe urinary tract abnormalities, and bilateral cryptorchidism in males, as seen in our patient. PBS has an incidence of 2–4 cases per 100,000 births, being more common in males; rare cases have been reported in females, who lack gonadal anomalies. Despite various theories, the exact cause of Prune Belly syndrome remains unidentified, and its genetic basis is unknown. The predominance in males suggests an X-linked recessive defect, though this inheritance pattern is contradicted by rare female cases and untreated affected males who experience azoospermia. PBS has also been associated with chromosomal anomalies, including trisomies 13, 18, and 21. A defect in normal mesodermal development has also been suggested, particularly in the intermediate and lateral plate mesoderm, which would affect abdominal musculature, mesonephric and paramesonephric ducts, and urinary organs. Possible mechanisms include HFN1B gene deletion, hemizygous missense mutations in the X-linked Filamin A gene, and CHRM3 loss-of-function mutation.

PBS diagnosis is clinical, typically identified by the characteristic triad at birth. Prenatal diagnosis may be possible via routine ultrasound between 20 and 30 weeks, where conditions like bilateral hydronephrosis and bladder dilation may be observed, as in our case. In severe obstructive uropathy, prenatal intervention may include vesicoamniotic shunting to prevent otherwise fatal neonatal complications, mainly pulmonary hypoplasia. Postnatal treatment focuses on improving renal and pulmonary function, with variable impacts on the child's health and quality of life. Surgical needs are classified into three categories based on patient needs: urinary tract reconstruction, abdominal wall repair, and orchiopexy. A multidisciplinary hospital evaluation by urology and pediatric surgery determined our patient did not require immediate surgical intervention.

PBS survival rates have improved significantly in recent years thanks to early diagnosis and management, although there is still an approximate 30% mortality rate, primarily due to perinatal pulmonary failure. Our patient had no pulmonary complications, and a transthoracic echocardiogram ruled out cardiac anomalies. The patient was discharged with outpatient follow-up by urology, nephrology, and pediatric surgery for long-term prognosis optimization.

5. Conclusion

Prune Belly syndrome is a rare congenital anomaly characterized by abdominal wall hypoplasia, bilateral cryptorchidism, and urinary tract abnormalities, posing a diagnostic and therapeutic challenge, especially in resource-limited settings. Early identification and multidisciplinary management are essential for improving prognosis, as these patients often present with complications affecting multiple organ systems.

This case emphasizes the importance of prenatal diagnosis and conservative intervention, which allowed renal function

stabilization and avoided immediate surgical procedures in our patient. Despite the complexities and risks associated with PBS, a comprehensive approach and specialized follow-up can facilitate a favorable outcome and optimize the affected individuals' quality of life.

This report also highlights the need for continued research into the genetic basis and pathogenic mechanisms of PBS to develop new prevention and treatment strategies that improve outcomes for patients diagnosed with this rare condition.

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