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A Rare Presentation of Prune Belly Syndrome in a Female Neonate: Case report and review of literature

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ABSTRACT

Prune Belly syndrome (PBS) is a rare congenital anomaly of uncertain etiology almost exclusive to males. There are variants of Prune Belly syndrome which are characterised by partial or unilateral hypoplasia of the abdominal wall muscles and may be associated with other malformations. These variants are described as incomplete or Pseudo Prune-Belly syndromes (PPBS). We report and discuss the case of a preterm female newborn, in whom clinical examination revealed deficient abdominal muscle, and she also had a soft systolic ejection murmur loudest over the pulmonic area. Abdominal ultrasound scan demonstrated no urinary tract anomalies; the presence of atria septal defect was confirmed on echocardiogram, and she had normal kidney function test results. She was referred to the paediatric surgical department and had follow-up cardiologist visits.

Keywords: Pseudo prune belly syndrome; Abdominal wall laxity; Atria Septal Defect; Female; Neonate

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Introduction

Prune Belly syndrome (PBS) is a rare congenital disorder with an incidence of 3.8 cases per 100,000 live births in males and 1.1 per 100,000 in females (Druschel 1995; Routh et al. 2010). This syndrome consists of a classical triad of deficient development of abdominal muscles that causes the skin of the abdomen to wrinkle like a prune, bilateral cryptorchidism, and abnormalities of the urinary tract such as bilateral gross hydronephrosis, megaureter and megacystitis (Tagore et al. 2011). There are variants of Prune Belly syndrome which are characterised by partial or unilateral hypoplasia of the abdominal wall muscles, and may be associated with ipsilateral renal, testicular or osteo-articular malformations. Other systemic involvement could include pulmonary, skeletal, gastrointestinal or cardiovascular malformations (Zugor et al. 2012; Grover et al. 2017). These variants are described as incomplete or Pseudo Prune-Belly syndromes (PPBS) and appear to be more deleterious due to a higher incidence of urethral atresia, especially in females (Zugor et al. 2012; Grover et al. 2017). We present a case of Pseudo Prune-Belly syndromes in a female neonate associated with a congenital heart disease.

Case presentation

This is the case of an 8-hour-old female neonate of non-consanguineous parents; that was referred to our facility with complaints of congenital abdominal wall malformation. She was delivered via spontaneous vaginal delivery at 36 weeks' gestation and had good extra-uterine transition with a birth weight of 2,070 grams. Her mother was regular with her antenatal clinic visits, received only prescribed medications and had unremarkable laboratory results. She reported a positive history of febrile illness without rashes during early pregnancy, but there was no exposure to ionising radiation, no history of chronic pathology, and no notion suggestive of oligohydramnios.

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On examination, she was afebrile, not pale, acyanosed with no dysmorphic features. Abdominal examination revealed a loose abdomen with thin skin and visible peristalsis [Figure 1]. Her intestines could be felt beneath her skin on abdominal palpation. The external genitalia were feminine; there was no sexual differentiation anomaly; anus was present and permeable. Cardiovascular system examination revealed normal peripheral arterial pulses, with capillary refill time of <3 seconds and oxygen saturation of 97% in room air. The auscultatory findings included a normal heart rate, soft systolic ejection murmur loudest over the left upper sternal border (pulmonic area), combined with a wide, fixed S2 splitting. She was not dyspnoiec on respiratory examination, with normal respiratory rate and breath sounds. Her neurological assessment also noted normal findings.

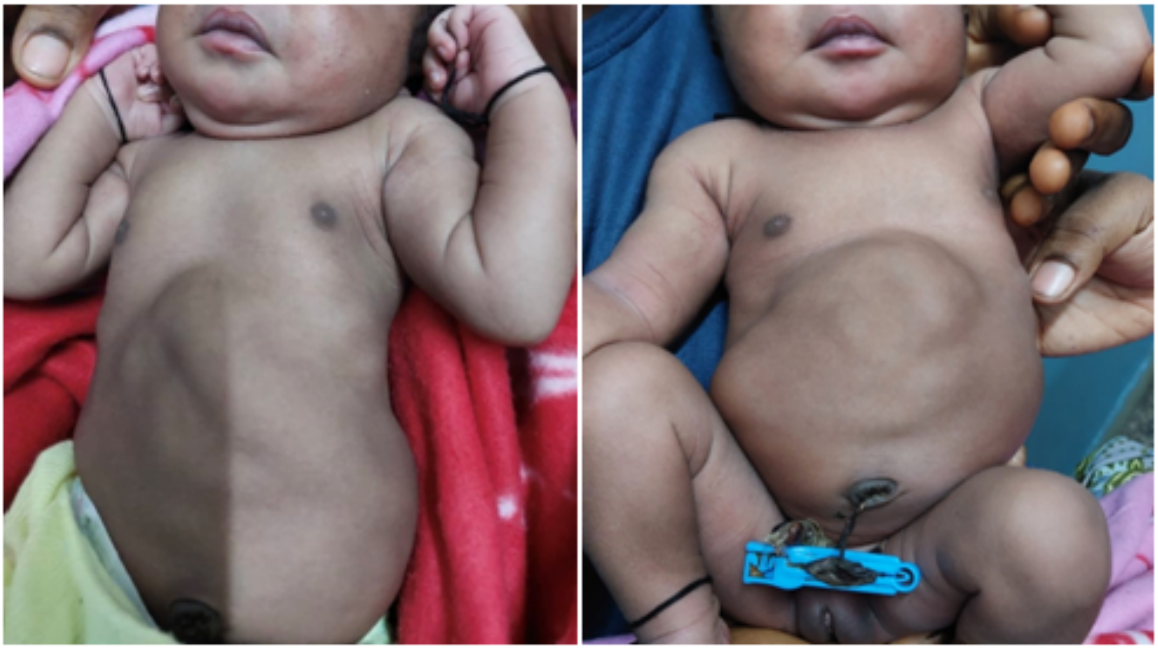

Figure 1: A female neonate having a loose abdomen with thin skin and visible peristalsis

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Initial blood investigations revealed normal electrolytes, urea and creatinine levels; abdominal ultrasound showed a diffusely thin abdominal wall which measured 0.34cm in thickness. Both kidneys were normal in size and showed good corticomedullary differentiation and normal renal sinus. There were no calculi, hydronephrosis, or megaureter. A transthoracic echocardiogram revealed an isolated secundum Atrial Septal Defect of 4mm with left to right shunt, and normal biventricular function.

Having an incomplete triad of prune belly syndrome, her final diagnosis was Pseudo Prune-Belly syndrome with asymptomatic Atrial Septal Defect. She received conservative care and remained clinically and haemodynamically stable. She is tolerating breastfeeding well at the time of referral to the paediatric surgical department, and cardiologist follow-up visits were continued.

Discussion

The Prune-Belly syndrome was first described by Fröhlich in 1839 (Frölich F. 1839). It is a rare congenital disorder with an incidence of 3.8 cases per 100,000 live births in males and 1.1 per 100,000 in females (Druschel 1995; Routh et al. 2010). The real etiology of the PBS is yet to be identified, but three plausible hypotheses have however been suggested, including the urethral obstruction malformation complex hypothesis (Greskovich et al. 1988) which proposes that pressure atrophy of the abdominal wall muscles occurs when urethral obstruction leads to massive distension of the bladder and ureters. Bladder distension would also interfere with descent of the testes and thus be responsible for the bilateral cryptorchidism. According to mesodermal defect hypothesis (Moore 1988) aberrant development of the derivatives of the first lumbar myotome between 6 and 10 weeks of gestation leads to a patchy muscular deficiency or hypoplasia of the abdominal wall as well as to urinary tract abnormalities. A third hypothesis is the yolk

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sac theory, which proposes a dysgenesis of the yolk sac and allantois as the basis of the PBS (Straub et al. 1981). PBS is predominant in male infants with a prevalence of more than 95% (Ramasamy et al. 2005). Based on the sex predominance, a genetic influence has eventually been evoked with a possible transmission through autosomal recessive sex-linked genes (Ramasamy et al. 2005).

Prune-Belly syndrome consists of a classical triad of deficient development of abdominal muscles that causes the skin of the abdomen to wrinkle like a prune, bilateral cryptorchidism, and abnormalities of the urinary tract such as bilateral gross hydronephrosis, megaureter and megacystitis (Tagore et al. 2011). Females present with abdominal wall deficiency and a dysmorphic urinary tract without any associated gonadal anomaly (Oliván-Gonzalvo 2021). PBS may also present with a wide range of severity, distinguishing three major categories of presentation as described by Woodard in 1985 [Table 1] (Woodard 1985). Category III, also called pseudo-PBS, includes patients with mild triad features or incomplete forms, and whose renal function is normal or mildly impaired as seen in our patient who also had atrial septal defect. Routh et al reports other malformations that may be associated with PPBS, notably pulmonary, cardiac, skeletal, gastrointestinal or genital malformations, with significant incidences of 25% for cardiovascular defects, 24% for gastrointestinal birth defects, 23% for musculo-skeletal defects, 58% for respiratory defects and 15% for genital malformations (Straub et al. 1981; Routh et al. 2010; Fette 2015).

Therefore, the holistic management of patients with PBS requires a multidisciplinary collaboration including the urologist, paediatrician, genetician, nephrologist, pediatric surgeon and anesthesiologist. The main surgical interventions consist of a series of operations including the reconstruction of the urinary tract, abdominoplasty and orchidopexy (Ngwanou et al. 2020). In females

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with pseudo-PBS having no urinary tract anomalies, abdominal wall reconstruction is necessary as the abdominal muscles deficiency is not only an aesthetic problem. It can also predispose her to gross motor development delayed, because it does not allow the level of force required to maintain balance and stabilisation of the spine when performing various activities such as sitting, walking or running (Arlen et al. 2016; Pomajzl et al. 2020). In addition, our patient has an ASD of 4mm, and patients with ASDs smaller than 5 mm often experience spontaneous closure of the defect within the first year of life and may not require intervention (Behjati-Ardakani et al. 2016). However, regular cardiologist reviews with echocardiogram monitoring is recommended.

Table I: Classification of Prune Belly Syndrome

Category	Characteristics
I	Renal dysplasia Oligohydramnios Pulmonary hypoplasia Potter's facies Urethral atresia
II	Full triad features Minimal or unilateral renal dysplasia No pulmonary hypoplasia May progress to renal failure
III	Incomplete or mild triad features Mild to moderate uropathy No renal dysplasia Stable renal function No pulmonary hypoplasia

Conclusion

We presented a case of pseudo prune belly syndrome with atrial septal defect in a female neonate. Early detection is vital for appropriate management and timely surgical interventions.

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References

1. Arlen AM, Kirsch SS, Seidel NE, Garcia-Roig M, Smith EA, Kirsch AJ (2016). Health-related Quality of Life in Children With Prune-belly Syndrome and Their Caregivers. Urology.87:224-7. doi: 10.1016/j.urology.
2. Behjati-Ardakani M, Golshan M, Akhavan-Karbasi S, Hosseini SM, Behjati-Ardakani MA, Sarebanhassanabadi M (2016). The Clinical Course of Patients with Atrial Septal Defacts. Iran J Pediatr.26(4):e4649
3. Druschel CM (1995). A descriptive study of prune belly in New York State, 1983 to 1989. *Arch Pediatr Adolesc Med*.149(1):70-76.
4. Fette A (2015). Associated rare anomalies in prune belly syndrome: a case report. J of Ped Surg Case rep.3(2):65-71.
5. Frölich F (1839). Der Mangel an Muskeln, inshesondere der Seitenbauchmuskulatur. Inaug Diss.1
6. Greskovich FJ, Nyberg LM (1988). The Prune Belly syndrome: A review of its etiology, defects, treatment and prognosis. J Urol.140:707-12.
7. Grover H, Sethi S, Garg J, Ahluwalia A (2017). Pseudo Prune Belly syndrome: diagnosis revealed by imaging: a case report and brief review. Pol J Radiol.82:252-7.
8. Moore KL (1988). The Developing Human. Clinically Oriented Embryology. Philadelphia, USA: W. B. Saunders Company.
9. Ngwanou DH, Ngantchet E, Moyo GPK (2020). Prune-Belly syndrome, a rare case presentation in neonatology: about one case in Yaounde, Cameroon. Pan African Medical Journal.102(36):1-5
10. Oliván-Gonzalvo G (2021). Pseudo prune belly syndrome: a female case report. Iberoam J Med.3(1):98-99. doi: 10.5281/zenodo.4411082.
11. Pomajzl AJ, Sankararaman S (2020). Prune Belly Syndrome. In: StatPearls Treasure Island (FL): StatPearls Publishing.
12. Ramasamy R, Haviland M, Woodard J, Barone J (2005). Patterns of inheritance in familial Prune Belly syndrome. Urology.65(6):1227.
13. Routh JC, Huang L, Retik AB, Nelson CP (2010). Contemporary epidemiology and characterization of newborn males with prune belly syndrome. *Urology*.76(1):44-48.
14. Straub E, Jurgen S (1981). Etiology and pathogenesis of the prune belly syndrome. Kidney Int.20(6):695-9.
15. Tagore KR, Ramineni AK, Vij aya Lakshmi AR, Bhavani N (2011). Prune Belly syndrome. Case Rep Pediatr:1-3.

©SLJBR Volume 16. Number 2. December 2025 ISSN (Print), ISSN (Online First)

16. Woodard JR (1985). Prune Belly syndrome. In: Edited by King LR, Kelalis PP, Belman AB. Clinical Pediatric Urology. Philadelphia: WB Saunders;p. 805-24.

17. Zugor V, Schott E, Labanaris P (2012). The Prune Belly syndrome: urological aspects and long-term outcomes of a rare disease. Pediatr Rep.4(2).