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Pseudo Prune Belly Syndrome, rare case report

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Title page

Pseudo Prune Belly Syndrome, rare case report

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Key clinical message

Even though the pseudo prune syndrome occurs occur at the same incidence as Prune Belly Syndrome it has given less emphasis and goes unrecognized. Emphasis should be given with patients with any of the triad and thorough clinical examination and imaging is needed for early diagnosis and management

Pseudo prune Belly Syndrome (PPBS) is a term used to describe infants who do not fulfill the criteria of classic PBS, consisting of abdominal wall muscle deficiency or hypoplasia, urologic abnormality, and cryptorchidism. So far there are only few reports. To best of our knowledge there is no case report of Pseudo Prune Belly Syndrome in Ethiopia. Here we present a case of 45 days old female infant Pseudo prune Belly Syndrome

Key terms : Infant, Prune Belly Syndrome, Pseudo Prune Belly Syndrome, Case report, Ethiopia

Introduction

Prune belly syndrome (PBS), also named Eagle-Barrett syndrome, is a rare multisystem congenital disorder with a triad presentation of urinary tract dilation, deficient abdominal wall musculature and bilateral undescended testes (1–3). There is a broad spectrum of severity of the condition, with some patients not surviving the neonatal period to others with minimal abnormalities. PBS affects 3.8 cases per 100,000 live births and is more common in infants of African descent (3). The male-to-female ratio is reported to be 5:1 and females with PBS have abdominal wall muscle deficiency and urinary tract dilation, but no gonadal anomalies (3,4). The etiology of PBS is unknown although no specific gene has been identified for PBS, certain genes have emerged as candidates HNF1β is a transcription factor and *CHRM3,* which encodes the M3 muscarinic acetylcholine receptor. Most cases of PBS occur sporadically. Currently, there are no large cohort investigations regarding the genetics of PBS, and therefore, the genetic basis for PBS remains unknown (1,3). It is estimated to affect 1 in 30,000-40000 live births worldwide with more than 90% of the cases occurring in boys (2,5).

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Children with prune belly syndrome presents with renal, ureteral and urethral abnormalities with complications ranging from urinary tract infections to chronic kidney disease, lack of abdominal muscles leads to a poor cough mechanism which in turn leads to increased pulmonary secretions. The weak abdominal wall muscles also lead to constipation (5–7). The prognosis may vary from stillbirth to a normal life expectancy depending on the degree of pulmonary and renal compromise; mortality is 20% within first month and 50% within 2 years (5).

The term "pseudo prune belly" has been used to describe a patient with normal abdominal wall, absent or incomplete cryptorchidism and urinary tract like that seen with PBS. Moreover, one might expect that for this group of patients, involvement of urinary tract and the clinical course will be less severe than that for patients with PBS (1).

Case History/examination

The index case was 45 days old male infant born to para-III mother delivered at home with no antenatal care follow up, historically presented with right side abdominal distension since birth. Otherwise the infant was fine and no other complaint. This patient vital sign and anthropometry was normal for age. On clinical examination the abdomen is distended and soft on the right side but normal on left side (Fig.1). For this patient presentation abdominal ultrasound was done at our hospital.

On ultrasound left side abdominal wall muscles (both rectus abdominis and oblique muscles) were visualized with normal thickness and echotexture (Fig.2). Right side of midline no abdominal wall muscles visualized (Fig.2). Bowel loops wall was visualized just beneath the abdominal wall subcutaneous tissue. On further assessment only left testis was visualized in scrotal sac with minimal surrounding fluid. Right scrotal sac was not developed and the right testis was not visualized in inguinoscrotal region (Fig.3A). There was 9mm by 7mm measuring oval shaped structure seen in right side of retroperitoneum anterior to right psoas muscle just below the renal hilum (Fig.3B). The oval shaped structure has similar shape and echotexture to left side intrascrotal testis. Bilateral kidneys are located in normal position with no evidence of hydronephrosis (Fig 4A).The urinary bladder was minimally distended with urine (estimated volume of 20cc) with normal wall thickness (Fig.4B).

Differential diagnosis

So on imaging the diagnosis of pseudo prune belly syndrome consistent right side abdominal wall muscles deficiency with ipsilateral retroperitoneal testis was made.

Outcome and follow up

Since the infant was stable he was put on follow up for future surgery for right testis and to follow left side hydrocele.

DISCUSSION

The incidence of PBS is estimated to occur in 1:40,000 live births with more than 95% affecting males, its characterized by triad of deficient abdominal wall muscles, bilateral cryptorchidism and abnormalities of urinary tract such as hydronephrosis, hydroureter/mega ureter and persistent urachus, megacystis with megalourethra (1,2). Literature on the cause and embryogenetic factors of PPBS and PBS is controversial, majority postulates that its due to three possible factors occurring during embryogenesis; like severe bladder outlet obstruction, dysgenesis of yolk sac and possibly due to abdominal muscle deficiency secondary to migrational defect of mesoblast during early 1sttrimester of pregnancy (1,2,8). Prognosis of infants with PBS is poor with IUFD, still births and early infant death being common (5,9,10). Physical finding that support the diagnosis of PBS include a penile abnormalities (megalourethra, urethral atresia), patent urachus and musculoskeletal abnormalities (1,2,4,7). The clinical severity of PBS is widely variable, depending upon the timing, location, and degree of the embryologic insult. Some patients have severe manifestations which lead to still birth or infant death while others demonstrate very mild disease which requires little or no therapy (6,9,11).

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The terminology of "Pseudo Prune Belly Syndrome (PPBS) has been Suggested to define those who do not complete the triad of PBS (1,4,12). PPBS is a term used to describe infants who do not fulfill the criteria of classic PBS, consisting of abdominal wall muscle deficiency or hypoplasia, urologic abnormality, and cryptorchidism, terms such as "incomplete prune belly syndrome" or "partial prune belly Syndrome" have been used to describe children cryptorchidism, mild urinary tract abnormalities and mild or unilateral deficiency of abdominal musculature: deficient prune like abdominal wall and normal genitourinary system. Numerous report was described in children with only mild diastasis of the rectus abdominis musculature or abdominal wall deficiency with normal urinary tract imaging that the syndrome goes unrecognized (8,11). The term "pseudo prune" or partial prune can then be applied to a patient once the urologic hallmarks of PBS are recognized on urologic imaging that is done for indications other than PBS (1). PPBS includes patients who exhibit: (1) unilateral abdominal wall muscular deficiency, cryptorchidism, and urinary tract anomalies; (2) abdominal wall muscular deficiency alone; (3) abdominal muscular deficiency with either urinary tract anomalies or cryptorchidism; or (4) characteristic urinary tract anomalies and cryptorchidism but normal abdominal wall musculature (3,4). Sometimes, abdominal wall muscular hypoplasia is subtle or limited to diastasis of the rectus abdominis muscles (4).

Although the incomplete expression of the classic triad illustrated by our case is stated to occur at the same incidence as the complete triad, PPBS has not been emphasized in the literature (3,4). PPBS is generally characterized by partial or unilateral abdominal wall deficiency or unilateral undescended testis that can usually be palpable in the inguinal canal (4,11). Our patient is case of PPBS, which can be considered as a possible variant of the syndrome.

Conclusion

Even though the pseudo prune syndrome occurs occur at the same incidence as Prune Belly Syndrome it has given less emphasis and goes unrecognized. Emphasis should be given with patients with any of the triad and thorough clinical examination and imaging is needed for early diagnosis and management. Further research is recommended for better understanding of the disease and to determine the optimal management.

Author's contribution

Rabirra Waktola: Conceptualization, Data curation, Visualization, Writing - original draft, Writing - review & editing Tajudin Adem: Data curation, Resources, Writing - original draft, Writing - review & editing, Tesfaye Negasa: Writing - original draft, Merga Daba: Supervision, Validation

Declaration of patient consent

The written informed consent was taken from patient's parents. They gave written informed consent to use the clinical information, laboratory data and radiologic images to be used for the publication.

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Conflict of interests

There is no conflict of interests

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