



PRUNE BELLY SYNDROME: A CASE REPORT ASSOCIATED WITH AUTISM AND HYDROCEPHALY

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<p>Article Info</p> <p><i>Received 15/09/2015</i> <i>Revised 27/09/2015</i> <i>Accepted 12/10/2015</i></p> <p>Key words: Prune-belly syndrome, autism, hydrocephaly, Eagle-Barrett syndrome</p>	<p>ABSTRACT</p> <p>Prune-belly syndrome (PBS) consists of a triad of deficient or absent abdominal wall musculature, abnormalities of the urinary system and bilateral intra-abdominal testes. Several associated conditions have been reported, including pulmonary (58%), cardiovascular (25%), gastrointestinal (24%) and musculoskeletal (25%). This case report describes a 5-year-old male PBS patient with hydrocephaly submitted to ventriculoperitoneal shunt. He was also diagnosed with autism at age two. Although PBS patients have classic clinical features, detailed investigation and careful follow up are mandatory in order to evaluate the whole context of this syndrome in view of the variety of organ involvement.</p>
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INTRODUCTION

Prune-belly syndrome (PBS), also known as Eagle-Barrett syndrome consists of a triad of deficient or absent abdominal wall musculature, abnormalities of the urinary system and bilateral intra-abdominal testes. Approximately 96% of PBS patients are males, while females are rarely affected [1-3]. The great majority of those affected are stillborn or die within the first week of life as a result of pulmonary or renal complications. The prevalence of PBS is estimated to be 3.76/100.000 of live births for those who survive. About 43% of PBS patients are born prematurely [4]. Several associated conditions have been reported, including pulmonary (58%), cardiovascular (25%), gastrointestinal (24%) and musculoskeletal (25%), but there is no description regarding PBS, hydrocephaly and autism.

Case report

The patient is a 5-year-old male. He was born at 38 weeks, without fetal distress or asphyxia. His weight was 3065g. His head circumference was 32.5cm, his APGAR score were 7 and 9. His parents are non-consanguineous and he has an older healthy daughter.

There is no known family history of hereditary disorders nor other relevant history.

He was first evaluated by a pediatric neurologist when he was 5 months old due to a global developmental delay and macrocrania. He had flaccid, wrinkled abdominal skin whose clinical features were consistent with prune-belly syndrome (PBS) (Figure 1), but no post-natal interventions were done. His neurological exam demonstrated a bulging fontanel, with his head circumference measuring 45.5 cm.

A cranial CT was immediately requested, confirming the diagnosis of hydrocephaly. He was admitted to the hospital, where a neurosurgeon performed a ventriculoperitoneal shunt. (Figure 2)

He was also referred to a pediatric surgeon due to cryptorchidism. A urinary ultrasound was performed and revealed class I cyst on his right kidney, but no other abnormalities (Figure 3)

At age 2 he began to demonstrate repetitive movements, however he did not attempt to make eye contact and did not yet pronounce a single word. He also demonstrated a lack on social-emotional reciprocity,



therefore, he met the DSM-V criteria for an autism spectrum disorder. He was also very irritable and shouted much of the time. For this reason he was medicated with Risperidone 0.5mg a day, with some improvements in his behavior. Currently he attends in a multidisciplinary center.

DISCUSSION

A great number of pediatricians will encounter at least once a patient with PBS in their clinical practice. It is important to have the knowledge to recognize it and have the knowledge about this syndrome and its associated conditions. PBS presents with classic clinical and imaging features. The classic triad encompasses [1] a deficient abdominal wall, [2] urinary tract abnormalities and [3] cryptorchidism. The incidence in male infants is cited as 1.1/100.000. Epidemiologic information is not available in Brazil, but data from the United States indicates that the Afro-Caribbean population is most affected. There also appears to be an increased incidence of PBS in children from young mothers [4].

About 43% of PBS patients are born prematurely, and a vast majority die within the first week of life as a result of pulmonary or renal complications. Those who survive have varying degrees of hydroureteronephrosis with poor muscular function of the ureters and bladder. Chronic renal insufficiency or failure and recurrent urinary tract infections are very common complications. There is also an increased risk of urinary tract rupture with even relatively minor abdominal trauma [5].

Hassett identified many other associated conditions [1]. The most prevalent is pulmonary hypoplasia (58%), followed by cardiovascular (25%), gastrointestinal (24%) and musculoskeletal malformations (25%). Cardiovascular abnormalities includes PDA, Tetralogy of Fallot, atrial septal defect and VSD. Gastrointestinal features include malrotation in 40% of

cases, imperforate anus and omphalocele. Scoliosis, clubfeet and congenital hip dislocation are some examples of musculoskeletal findings. PBS can be associated with other syndromes, including Turner’s, Down’s, and trisomy 13 and 18[6].

Navarro-Arenas describes an association of PBS and schizencephaly [7]. A literature review did not yield any reports of PBS associated with hydrocephaly and autism. Fernell published a study in 1991 about behavioral problems and autism in children with hydrocephalus. Autistic features were present in 13% of these children [8].

Two major theories have been suggested for the etiology of PBS. Recent studies demonstrate haploinsufficiency of hepatocyte nuclear factor 1- beta, although inherited genetic mutations alone cannot explain the pathogenesis of PBS. The most accepted mechanism is pressure atrophy of the abdominal wall musculature due to urethral obstruction. Bladder and ureters distension also leads to cryptorchidism, interfering with descent of the testes [5]. Antenatal diagnosis can occur around the 2nd trimester [5]. The most common presentation of PBS is bilateral oligohydramnio, hydroureter, hydronephrosis and a distended, thin-walled bladder. All muscles layers are compromised below the umbilicus and replaced by fibrous tissue. Peripheral abdominal wall shows normal or nearly normal muscle and fascial layers [5].

Hassett proposed a classification according to the time of diagnosis and clinical manifestations [1] (Table 1). This case is classified as category 3, which includes normal renal function and mild degree of uropathy, although he had typical external features. Category 3 occurs in 40% of infants born with PBS [9]. Hydrouretheronephrosis and dilated trabeculated bladder may give a pseudo-prune belly appearance of the abdominal wall, and both should be considered in the differential diagnosis. Megacystis-microcolon syndrome should also be considered although it occurs predominantly in girls.

Table 1. PBS Classification

Category 1 (20%)	Category 2(40%)	Category 3 (40%)
Pulmonary hypoplasia and/or pneumothorax	Typical external features	Mild or incomplete external features
Olygohydramnio	Hydrouretheronephrosis	Normal renal function
Renal dysplasia	Uropathy	Mild degree of uropathy
Urethral obstruction patent urachus	Renal dysplasia	
Club feet	Risk of urosepsis	
	Risk of azotemia	

CONCLUSION

Absence or hypoplasia of the abdominal wall muscles is the main recognizable feature of PBS triad, however it is imperative to be aware of its related hidden malformations, like cryptorchidism, urinary and pulmonary anomalies. Usually, there are no neurologic or psychiatric manifestations, but this case reports an unusual clinical presentation of PBS, with hydrocephalus and autism. Autism is thought to be a secondary complication of hydrocephalus in this patient. It is possible that the

concurrence of hydrocephalus and PBS was a merely coincidence, since their embryology and pathogenesis are very different. Detailed investigation and careful follow up in patients with PBS are mandatory in order to evaluate the whole context of this syndrome in view of the variety of organ involvement.

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STATEMENT OF HUMAN AND ANIMAL RIGHTS

All procedures performed in human participants were in accordance with the ethical standards of the institutional research committee and with the 1964

Helsinki declaration and its later amendments or comparable ethical standards. This article does not contain any studies with animals performed by any of the authors.

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