Prune Belly Syndrome Associated with Full Spectrum of VACTERL in a New Born

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ABSTRACT

Prune belly syndrome (PBS) is a rare congenital anomaly of uncertain etiology. Many associations of PBS with other malformations were previously reported, but only few cases of the association with VACTERL have been described. We report a rare case of a Moroccan new born with PBS and complete VACTERL association. The cause of this association is still unknown, but a common etiology is possible, especially when for the two syndromes, a defect in mesodermal differentiation, in early first trimester, has been suggested.

Key words:
Esophageal atresia, new born, Prune belly syndrome, VACTERL association

INTRODUCTION

Prune belly syndrome (PBS) is a rare congenital anomaly described most often in male newborns.[1] Classically, it combines a complete or partial triad made of an abdominal muscle deficiency, bilateral cryptorchidism, and dilated urinary tract. The VACTERL association is a nonrandom pattern of defects occurring together and includes at least three of its features.[2] They are two rare conditions with no definite etiology, but a defect in mesodermal differentiation, in early first trimester, has been suggested for the two syndromes.[3] Their association in a child is extremely rare and, to our knowledge, only few cases have been reported. We present the case of a new born with PBS and complete VACTERL association, and we discuss the possibility of a common etiology.

CASE REPORT

A three-day-old full-term new born was referred to our pediatric hospital for management of his esophageal atresia (EA), associated to other numerous abnormalities. It was diagnosed after several episodes of coughing and cyanosis occurring after attempts of feeding. The baby, born vaginally, was non-consanguineous, with no similar cases in siblings. We did not find any notion of teratogen exposure or toxic intake during this nonfollowed pregnancy. On physical examination, he was dyspneic, slightly hypotonic and multiple malformations were objectified: anal atresia, esophageal atresia which was confirmed by a thoraco-abdominal X-ray showing coiling of the nasogastric tube in dilated upper esophageal pouch, with presence of air in stomach suggesting a tracheo-esophageal fistula. Ultrasound examination of the abdomen showed bilateral gross hydronephrosis with megaureter, and echocardiography objectified a ventricular septal defect. All these findings gather a complete spectrum of VACTERL association. This infant also found to have a distended abdomen with lax abdominal wall revealing intestinal peristalsis and palpable kidneys and bladder [Figures 1 and 2]. He also presented X-ray showing a thoracic hemivertebra [Figure 3] bilateral cryptorchidism. These data, in association with dilated urinary tract, give us the full spectrum of PBS. Karyotype of this new born was normal (46, XY). The parents refused the surgery and any medical treatment, and then the patient died after 12 days of hospitalization from septic shock with respiratory failure.

DISCUSSION

The incidence of PBS is 3.8 cases/100000 live births[4] and that of VACTERL is varying from 1/3500 to 1.6/10000.[5]

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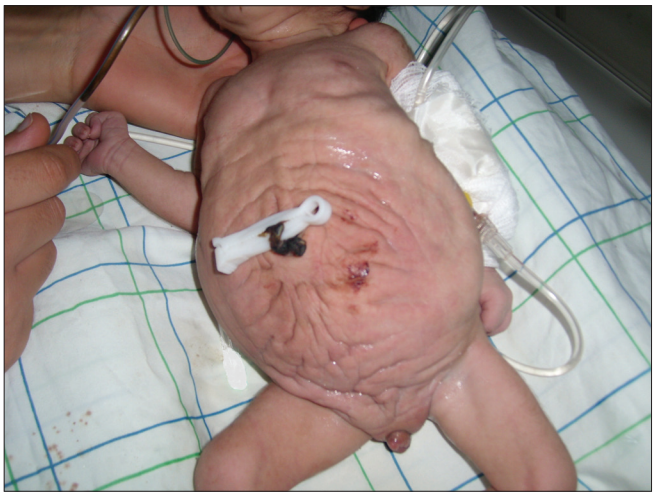

Figure 1: Image of our patient showing a distended abdomen, with a lax abdominal wall

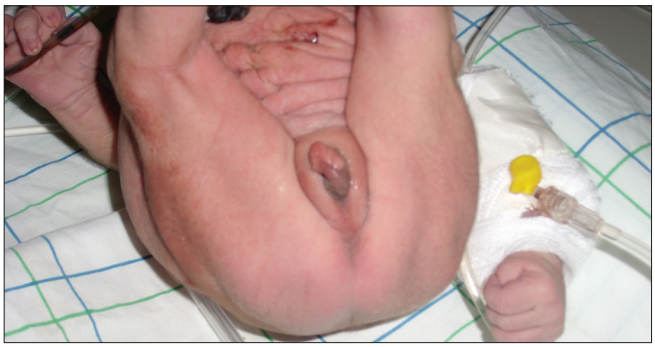

Figure 2: Anal atresia and cryptorchidism

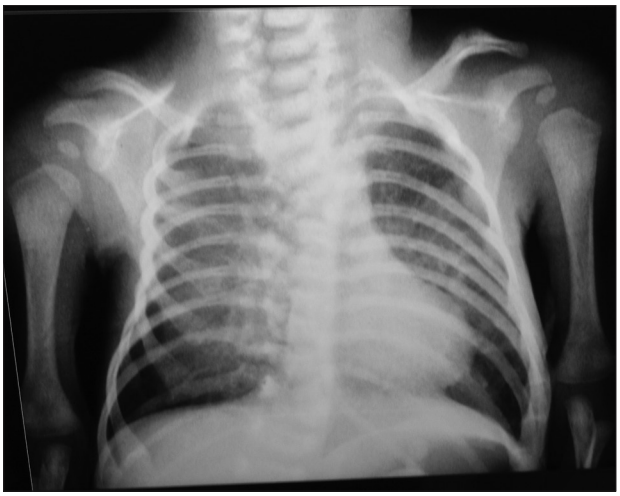

Figure 3: Chest-X-ray showing a thoracic hemivertebra

The incidence of the association of both conditions is not known. Internet research found only seven articles reporting such rare association.[6-10] Reinberg *et al*.[10] in 1993 reported a case of PBS, tracheo-esophageal fistula (TEF) associated with VATER syndrome with urethral atresia in a stillborn. Ozturk *et al*.[6] in 1994 reported concordance of complete PBS and VACTERL association in a premature male infant. Lukusa *et al*.[11] in 1996 described incomplete PBS in a female child with additional features of VACTERL association. Both the cases described by Ozturk and Lukusa did not have EA or TEF. Potter *et al*.[12] in 2002 reported a 33 weeks premature female neonate of PBS associated with TEF and urethral atresia. She underwent multiple operative procedures in antenatal as well in postnatal period and she died of overwhelming sepsis. Shah *et al*.[7] in 2004 reported the only case associating a full spectrum of VACTERL (all six components) with a complete PBS. It was about a term, 2.3 kg male neonate, who died on the third day of life without any surgical intervention. Another case has recently reported the presence of full spectrum of VACTERL (with esophageal atresia) in a female presenting a pseudoprune belly syndrome.[8] Our patient was a male which is confirmed by karyotype.

The cause and embryogenesis of PBS remain controversial. Three possible causes have been proposed: bladder outlet obstruction, mesodermal arrest, and dysgenesis of the yolk sac. Indeed, the second theory which postulates that the cause of PBS is a mesodermal arrest, occurring between the 6th and 10th weeks of gestation, provides also an explanation for the possible association of PBS with other malformations, especially with VACTERL.[3]

The prognosis of PBS is poor in our context. However, in developed countries, the prognosis usually depends on degree of renal dysfunction and pulmonary hypoplasia.[3] With contemporary care, the survival rate has improved in patients who survive the neonatal period, with approximately 25-30% of these patients experiencing chronic renal failure and end-stage renal failure.[3] In our case, the prognosis is poorest with the concordance of many severe malformations.

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