Prune belly syndrome associated with incomplete VACTERL

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ABSTRACT

A Prune Belly syndrome with VATER/VACTERL association is an extremely rare. They are either stillborn or die within few days of life, only few such cases have been reported in literature. We are presenting here a male neonate of Prune Belly syndrome associated with incomplete VACTERL with brief review of literature.

KEY WORDS: Esophageal atresia, Prune Belly syndrome, VACTERL, VATER

INTRODUCTION

Prune Belly syndrome (PBS) is also known as Eagle-Barrett syndrome, Osler-Parker syndrome, triad syndrome, abdominal muscles deficiency syndrome, mesenchymal dysplasia syndrome and consists of abdominal wall muscles deficiency or hypoplasia, urological anomalies and bilateral cryptorchidism. Esophageal atresia (EA) is estimated to occur in 1:2500 to 4500 live births and 50-70% of infants have associated anomalies. The incidence of 1.5% was reported for the full VATER association and 17.5% for the three components of the VACTERL association.

CASE REPORT

A 3 hours old 3 kg male neonate presented to us on March 31, 2006 with complaints of abdominal distension, absent anal opening, and frothing from mouth and nostrils since birth. He was a full term, normal hospital delivered first baby of a 22 years old mother. Antenatal history was not significant. Clinical examination revealed; laxity of distended abdominal wall, palpable kidneys and urinary bladder with bilateral undescended testes [Figure 1]. EA and imperforate anus were also present. Cardiovascular system examination revealed a pansystolic murmur. There were no bony/limb abnormalities detected clinically. Babygram revealed EA with distal fistula, fluid density, and little intestinal gas shadow in peritoneal cavity and sacral deformity [Figure 2]. Ultrasonography of abdomen confirmed bilateral gross hydroureteronephrosis [Figure 3] dilated and smooth-walled urinary bladder and testes were detected intra-abdominally. Perineal ultrasonography revealed a pouch perineal distance of 16 mm. The nature of intrinsic cardiac defect/cardiac anomaly could not be detected as echocardiography was not done.

At thoracotomy; ligation of distal trecheo-esophageal...
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tract; but the association of PBS with VACTERL is a rare occurrence. Internet search revealed only five such cases in literature. Reinberg case of PBS, TEF associated with VATER syndrome with urethral atresia in a stillborn. Ozturk et al. in 1994 reported concordance of complete PBS and VACTERL association in a premature male infant. Lukusa et al. 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. 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 as postnatal period and she died of overwhelming sepsis. Shah et al. in 2004 reported a term, small for gestational age, 2.3 kg male neonate with full spectrum of VACTERL association in addition to triad of PBS. He died on the third day of life without any surgical intervention.

Although EA is associated with other anomalies in more than half of the cases and PBS is also associated with other anomalies in 65-75% of cases outside the urinary tract; but the association of PBS with VACTERL is a rare occurrence. Unfortunately none of above explains the entire components of PBS.

The etiology and embryogenesis of PBS is controversial. Various theories have been proposed to explain the clinical features of PBS and they are fetal outlet obstruction, theory of mesodermal arrest and yolk sac theory. Although EA is associated with other anomalies in more than half of the cases and PBS is also associated with other anomalies in 65-75% of cases outside the urinary tract; but the association of PBS with VACTERL is a rare occurrence. Internet search revealed only five such cases in literature. Reinberg et al. in 1993 reported a case of PBS, TEF associated with VATER syndrome with urethral atresia in a stillborn. Ozturk et al. in 1994 reported concordance of complete PBS and VACTERL association in a premature male infant. Lukusa et al. 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. 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 as postnatal period and she died of overwhelming sepsis. Shah et al. in 2004 reported a term, small for gestational age, 2.3 kg male neonate with full spectrum of VACTERL association in addition to triad of PBS. He died on the third day of life without any surgical intervention.

Our patient was full term, 3-hour old, male had full spectrum of PBS and he also had associated all the component of VACTERL (Vertebral/sacral anomalies, anorectal malformation, cardiac, TEF, and renal anomalies) except limb anomalies. He underwent successful surgery for TEF and colostomy for anorectal malformation, but died on third post-operative day.

Both VACTERL association and PBS have a common etiology of a defect in the differentiating mesoderm in early first trimester. We believed that the occurrence of these two is another addition to the axial mesodermal dysplasia spectrum.

The management of EA with or without TEF is a straight one either a primary repair or staging procedures. Management of PBS is more complex and should be treated on individual basis and consists of abdominoplasty, bilateral orchiopexy, and urinary tract reconstruction. Few patients of PBS with progressive renal dysplasia, reflux nephropathy, etc, leading to chronic renal failure may need renal transplantation.

**DISCUSSION**

The term PBS was given by Williams Osler in 1901 to the wrinkled, wizened, prune like appearance of abdominal wall. The terminology of ‘pseudo prune’ belly syndrome (PPBS) has been suggested and used to define females and males who do not have the complete triad of PBS. The incidence of PBS is estimated to occur in 1:35,000 to 50,000 live births and 3-5% of PBS are in females as PPBS.

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