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PRUNE BELLY SYNDROME IN NEONATES

Pediatrics

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ABSTRACT

This is a rare disorder characterised by the triad of abdominal wall defects, urinary tract dilatation, and cryptorchidism. The prognosis remains guarded even in the most sophisticated centres of neonatal care. We are presenting 7 cases of Prune Belly Syndrome with variable presentation in neonatal period.

KEYWORDS

Prune Belly Syndrome, Cryptorchidism, hydronephrosis

Introduction: This syndrome was described first by Frolich in 1839, whereas Osler named this condition as "Prune Belly Syndrome", in 1901. This is also known by the names of "Eagle Barrett syndrome" and the "triad syndrome" [1]. The incidence reported in a recent study from United States is 3.8 cases per 100,000 male live births [2]. The complete syndrome is applicable to male infants, as the definition involves the presence of cryptorchidism, which can not be applied to females. The male: female ratio has been reported to be 5:1 in some recent studies [3]. It has been reported to be more common among preterm newborns and more common with younger age of the mother [2]. Familial occurrences, with X-linked recessive inheritance, have also been described in some series [4], but the causative and a confirmative statement is difficult to make in view of the rare nature of the disease. In hospital mortality rate remains upto 30% [2], despite best neonatal care due to natural history of the disease. The most common organ involved are the lungs.

Materials and methods: A retrospective analysis of seven cases of neonatal prune belly syndrome, admitted in the department of Pediatric surgery was carried out from January 2010-december 2017. They are presented below.

Case report: It is a retrospective analysis of all cases of prune belly syndrome, admitted in the department of pediatric surgery in neonates in the last 5 years, from 2010-2015. We had 7 cases, all of them were males, delivered full term, by normal vaginal delivery, with birth weights between 2.5-3.5 kg. Out of 7 cases, antenatal ultrasound was available in 5 cases, which was suggestive of bilateral hydronephrotic kidneys. All the cases were males and had bilateral cryptorchidism and flabby protuberant abdomen due to abdominal wall muscle weakness. One patient had bilateral small sized kidneys without ureteral dilatation with urethral agenesis and patent urachus. Urethral catheterisation was tried but failed. Cystogram done through the urachus revealed normal bladder with no reflux. The child continued to pass urine through the urachus and succumbed to refractory sepsis on day 45 of life. Three other cases had bilateral gross hydronephrosis, anorectal malformations and bilateral congenital talipes equinovarus. Colostomy was done in 2 of these cases. The third of them was associated with pulmonary hypoplasia and expired (fig 1), the other two survived and are on follow up. The other three cases had cryptorchidism, distended abdomen, bilateral hydronephrosis and congenital talipes equinovarus (fig 2). Ultrasonography of the abdomen and micturating cystourethrogram (MCUG) revealed hydronephrosis. They are also on follow up.

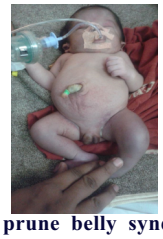

Fig 1: Neonate with prune belly syndrome and associated pulmonary hypoplasia.

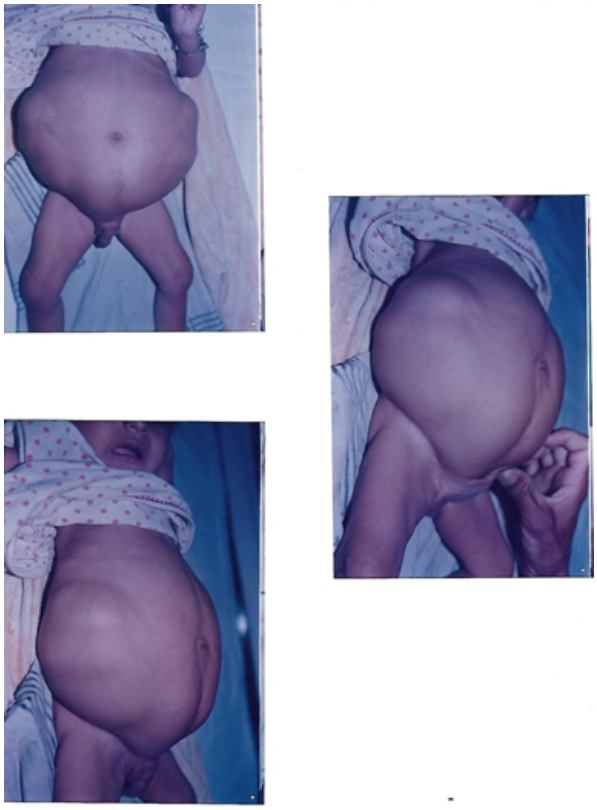

Fig 2: prune belly syndrome in a neonate with bilateral cryptorchidism and distended stomach.

Discussion: This syndrome is characterised by the triad of partial or complete absence of the abdominal wall, genitourinary abnormalities, including dilatation of the urinary collection system and bilateral cryptorchidism. There are 3 theories proposed to explain the pathogenesis of this condition. The first one includes transient bladder outlet obstruction, leading to megacystis, dilatation of the ureters, resulting in urinary ascites, pressure over the abdominal wall leading to deficiency of the abdominal wall and pushing up of the testes further into the abdomen which results in bilateral cryptorchidism. The second theory is that of mesodermal arrest. The arrest in development of the mesodermal somites leads to maldevelopment of the abdominal wall muscles and the genitourinary system. This theory also explains the association of this syndrome with other conditions, like vertebral, limb anomalies, cardiac and musculoskeletal defects, as described in certain case reports [5]. Also, the differential manifestations in monozygotic twins has been postulated to be due to unequal division of the embryo, leaving the affected twin with insufficient mesenchyme for the development of abdominal wall and genitourinary tract and

differential manifestations [6]. Laborie et al have postulated the lack of methylation at 6q24 locus, in affected monozygotic twin, leading to generalized maternal hypomethylation syndrome, responsible both for neonatal diabetes mellitus and PBS [7]. Deletion of hepatocyte nuclear factor-1 beta has also been described in some studies as causative factor [8].

The associated comorbidities include pulmonary hypoplasia, which is the main cause of mortality in this syndrome. They also have recurrent aspiration and resultant pneumonias due to poor abdominal wall strength. Gut malrotations are found in around 30% of the cases [9]. They also have intestinal atresias, stenosis, volvulus, imperforate anus, persistent cloaca. Musculoskeletal manifestations include congenital talipes equinovarus, dislocation of hip, limb and vertebral anomalies, pectus excavatum and pectus carinatum. The cardiac defects include atrial and ventricular septal defects, patent ductus arteriosus, tetralogy of Fallot.

The prognosis of the condition depends on the severity of the underlying lung and renal function abnormalities. The mortality rates have been reported to be upto 30% in various case series [2]. Management involves multispeciality approach, involving pediatric surgeons, plastic surgeons, psychiatrists, urologists, orthopedicians, and pediatricians. The surgical management involves reconstruction of abdominal wall in cases of recurrent pulmonary infections. The testes need to be managed by orchiopexy. The associated gastrointestinal, cardiac and musculoskeletal conditions might be needed to be addressed at appropriate times, when required. Though, the surgical interventions required during the neonatal period are few [2]. Those mostly reported are circumcision, urinary tract diversion and vesicostomy.

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