Caudal Duplication Syndrome—Report of a Case and Review of Literature

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Abstract A 3-day-old male neonate presented with features of anorectal malformation and duplication of the external genitalia. He was subsequently diagnosed with complete duplication of the colon, rectum, bladder, and urethra associated with spinal lipoma. We report this case of caudal duplication syndrome, considering its rarity and the diverse combination of gastrointestinal, genitourinary, spinal, and limb anomalies.

Keywords Caudal duplication · Anorectal malformation · Lipoma

Introduction

Caudal duplication syndrome is a rare entity that includes a wide spectrum of anomalies of the gastrointestinal, genitourinary, neurological, and vertebroskeletal systems. This entity was first described by Dominguez et al. [1] in 1993. Since then, few other cases have been reported in English literature. We report here a case of caudal duplication and review available literature with particular emphasis on the spectrum of the disease and its etiology.

Case Report

A 3-day-old neonate, first born male out of non-consanguineous marriage, presented with non-passage of

stool, abdominal distension, and bilious vomiting since birth. The child was delivered by normal labor at term and had cried soon after birth. There was no history of dribbling of urine and urinary retention. On examination, there were two hemi-phalluses with two patent external urethral orifices and two stenotic anal orifices—one orifice at the normal site and the other orifice to its left (Fig. 1). The lower limbs were normal. There was no overt spinal anomaly. The vitals were stable. There was no similar family history. There was no history of exposure to any drugs in the mother. A provisional diagnosis of anorectal malformation with duplication of the external genitalia was made. Exploratory laparotomy was done on 11th day of birth, which revealed complete duplication of the colon from the level of ascending colon up to the rectum and anus. There was only one cecum. Colostomy was done and four stomas were made (one pair for each colon) on the left side of the abdomen (Fig. 2).

The colostomy stomas functioned well and the child symptoms were relieved. Ultrasound and magnetic resonance imaging (MRI) of the abdomen were then done, which revealed duplication of the bladder with a mid-line sagittal septum (Fig. 3). Voiding cystourethrography (VCUG) showed complete duplication of the bladder in the sagittal plane and complete duplication of the urethra with equal caliber (Fig. 4). There was no vesicourethral reflux. There was no pubic diastasis. MRI of the abdomen also showed complete duplication of the colon from the level of ascending colon to the level of anal canal (Figs. 5 and 6). The kidneys, liver, spleen, aorta, and inferior vena cava were normal. Radiograph of the spine showed normal lumbosacral spine. There was no duplication of vertebra, agenesis, hemivertebra, and spina bifida. Echocardiography was normal. MRI of the spine revealed spinal lipoma (Fig. 7). The diagnosis

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Fig. 1 Perineum showing **a** two hemi-phalluses (*white arrows*) and **b** two stenotic anal orifices, one at normal site (*black arrow*) and the other (*white arrow*) to the left of it

Fig. 2 Four colostomy openings (*black arrows*) on the left side of the abdomen

of caudal duplication syndrome was made in this child with complete duplication of the colon, rectum, anus, bladder, urethra, and external genitalia associated with anorectal malformation and spinal lipoma.

At 1-year follow-up, the child has been doing well with normal functioning of the colostomy stomas. The developmental milestones are normal. The child has normal sensory and motor functions of the lower limbs. There are no urinary complaints. Staged correction of the anomalies of the intestine, bladder, urethra, and hemi-phalluses is planned.

Discussion

Caudal duplication syndrome was first described by Dominguez et al. [1] in 1993, in which there is varying

Fig. 3 **a** Ultrasound and **b** coronal MRI STIR images showing septum (*white arrows*) between the two urinary bladders (*UB*). MRI image also showing two urethras (*black arrows*)

Fig. 4 VCUG showing complete duplication of urinary bladder (asterisks) separated by the septum seen as linear defect (black arrows) with complete duplication of urethras (white arrows)

extent of duplication of structures arising from the cloaca and neural tube. It is a range of disease comprising gastrointestinal, genitourinary, vertebral, spinal, and limb

Fig. 5 Coronal T2-weighted MRI of the abdomen showing two ascending colons (black arrows), two descending colons (white arrows) and two urinary bladders (asterisks)

Fig. 6 Axial MRI STIR image showing two rectums (white arrows). Two urinary bladders (asterisks) and septum in between (black arrow) are seen anteriorly

abnormalities. Common gastrointestinal anomalies include duplication of the colon and rectum, which may be associated with a variety of other anomalies such as imperforate anus, rectal fistula, ventral hernia, omphalocele, duplication of terminal ileum, double appendices, Meckel's diverticulum, intestinal malrotation, and situs inversus [2]. Urogenital anomalies comprise duplication of the external and internal genitalia, ureters, and bladder and anomalies of the kidney [2]. Spinal anomalies include hemivertebra, sacral agenesis, myelomeningocele, diplomyelia, duplication of lumbar spine, butterfly vertebra, and spina bifida [2].

Exact etiology of caudal duplication syndrome is unknown. Various theories have been suggested. Incomplete separation of monozygotic twin has been postulated as the etiologic factor [3, 4] by Edwards [2]. Abnormal adherence between ectoderm and endoderm has been indicated as the cause, by Pang et al. [4, 5]. In view of Dominguez et al. [1], damage to caudal cell mass to the 23–25-day embryo is the causative factor. Bremer has proposed that pinching of the vesicle cavitating the solid intestinal tract at 6–7 weeks gives duplicate intestinal, genitourinary, and distal neural elements [2].

The patients present with a wide variety of symptoms or rarely may be asymptomatic [2]. Shah and Joshi [2] reported the case of an asymptomatic adult female with duplication of colon, rectum, anus, urinary bladder (UB), urethra, uterus, cervix, vagina, and external genitalia. Complete duplication of the urethra, aorta, and lower thoracic and lumbar vertebra with sacrococcygeal

Fig. 7 Coronal MRI T1-weighted (a) and STIR (b) images of the spine showing spinal lipoma (black arrows)

agenesis, spinal lipoma, and anorectal malformation in a female neonate was reported by Taneja et al. [4]. In a 2-year-old female child, a case of abnormal mass in the perineum with undeveloped feet, duplication of colon, external genitalia, and lumbosacral vertebra was reported by D'Costa et al. [3].

Treatment consists of staged correction of duplication anomalies [6]. Either stripping of mucosa or resection of duplicated colon and rectum is undertaken. Division of septum in the UB is done to make it a single chamber. Corrective surgery in the form of fusion is done for the external and internal genitalia. Any spinal anomaly is also corrected suitably. Many authors have reported near-normal cosmetic and functional result for these complicated anomalies. Shah and Joshi [2] suggested removal of one of the hemi-phalluses for cosmetic reasons in males. No corrective surgery was, however, done in the adult female patient reported by them. Liu et al. [6] reported the case of caudal duplication syndrome in which they did multiple stage correction. The bladder septum was removed, the two hemi-phalluses were fused to form one phallus, and the duplicated

colon was excised. The patient also had hydronephrotic left kidney and left megaureter, which were removed. Ultimate outcome is good [6].

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