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Caudal Duplication Syndrome: A Case Report of Challenging Imaging Findings and Management

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Abstract

Caudal duplication syndrome is a rare congenital disease caused by duplication of cloaca and notochord embryonic structures in various forms. The estimated prevalence of this syndrome is 1 per 100,000 births. A range of anomalies is observed in urinary and gastrointestinal tracts as well as in skeletal and neural structures of the spine and even limbs of these patients. Various factors such as genetic disorders and conjoined twinning have been mentioned in etiology of the disease. We introduce a case of this rare disease referred to our center for imaging as an infant with increased soft tissue in perineum without excretion of meconium from birth but with evidence of duplication in the genitourinary system (vagina and urethra) and gastrointestinal tract (rectum and colon) that was subject to reconstructive surgery.

Key Words: Caudal duplication syndrome, Congenital anomalies, Genitourinary duplication, Hindgut duplication, Pediatrics.

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1- INTRODUCTION

Caudal duplication syndrome (CDS) is a rare congenital disorder with a prevalence of ≤1 in 100,000 at birth that is associated with a wide range of anomalies and clinical manifestations (1). Duplication of the hindgut along with genitourinary abnormality is very uncommon (2).

Several hypotheses have been put forth to explain the etiology of CDS, including misexpression of homeobox (HOX) genes, an early insult to the urorectal septum, and other abnormal regression or duplication processes that disrupts the embryogenesis (3).

Imaging results are extensive in the study of these patients, play an important role in the diagnosis and determine the appropriate treatment approach as well as the surgical method.

2- CASE REPORT

The patient is a 4-day-old girl born by normal delivery. She was the first child of the family admitted to Akbar Pediatric Hospital of Mashhad University of Medical Sciences for intensive care. In postnatal examination, the increase of soft tissue in perineum (**Fig. 1**) was diagnosed as well as imperforate anus. There was no family history of congenital abnormalities and fetal screening had not been performed.

On physical examination, abdominal distension was observed due to the presence of imperforate anus. Other examinations showed normal results.

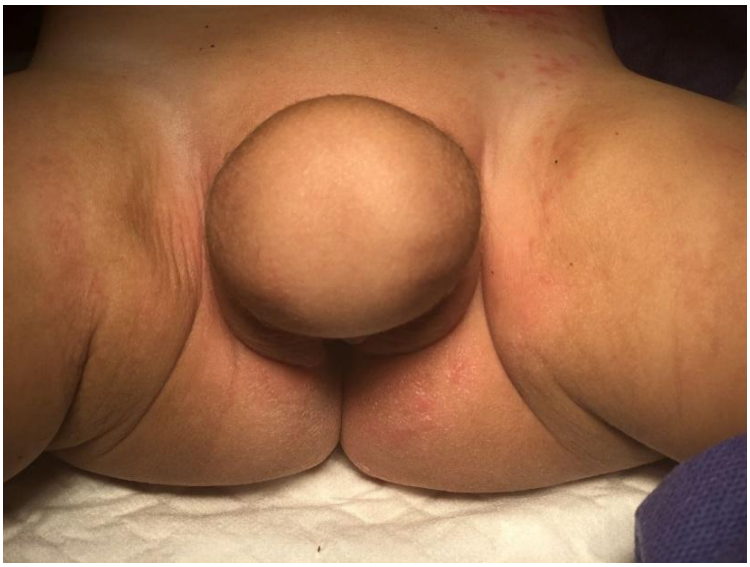


Fig. 1: Soft tissue augmentation area in the perineum of a three-day-old infant referred for imaging due to imperforate anus

For further examination, imaging modalities including conventional X-ray, echocardiography, abdominal and pelvic ultrasound, and pelvic MRI were performed at our center as shown in **Fig. 2** and **3**.

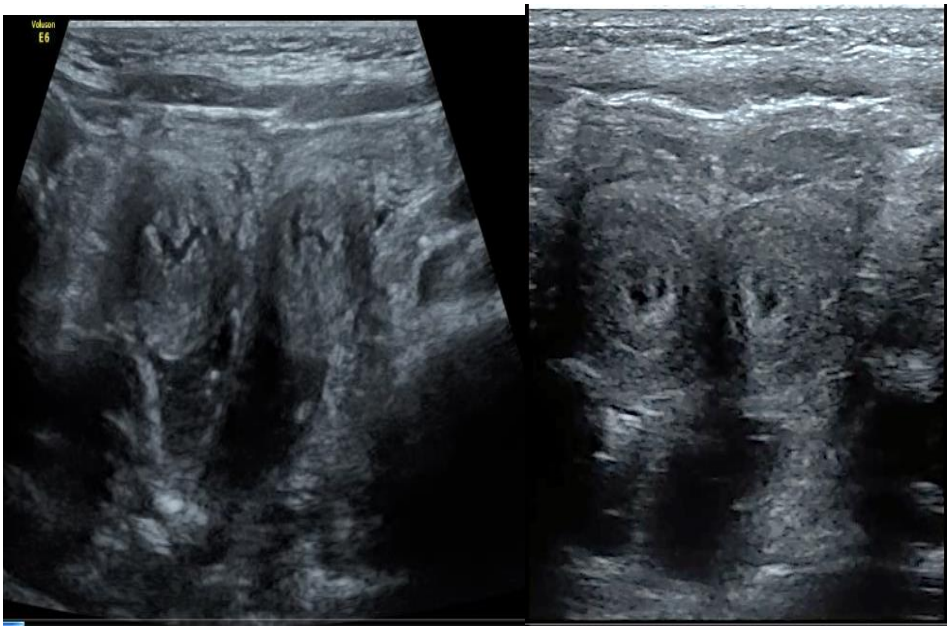
a) Conventional X-ray: Normal

Echocardiography: Normal

MRI: bladder duplication and uterus didelphys

Treatment course and surgical procedures:

Given the presence of an imperforate anus, an ostomy was created as a loop on the skin of the left side of the abdomen. Also, the soft tissue of the perineum was removed and the labia reconstructed.



(a) (b)

Fig. 2: a) Transverse ultrasound view with linear probe: image of two bladders in the anterior, two uteruses and two rectums in the posterior part **b)** Transverse ultrasound view with linear probe: image of two bladders in the anterior

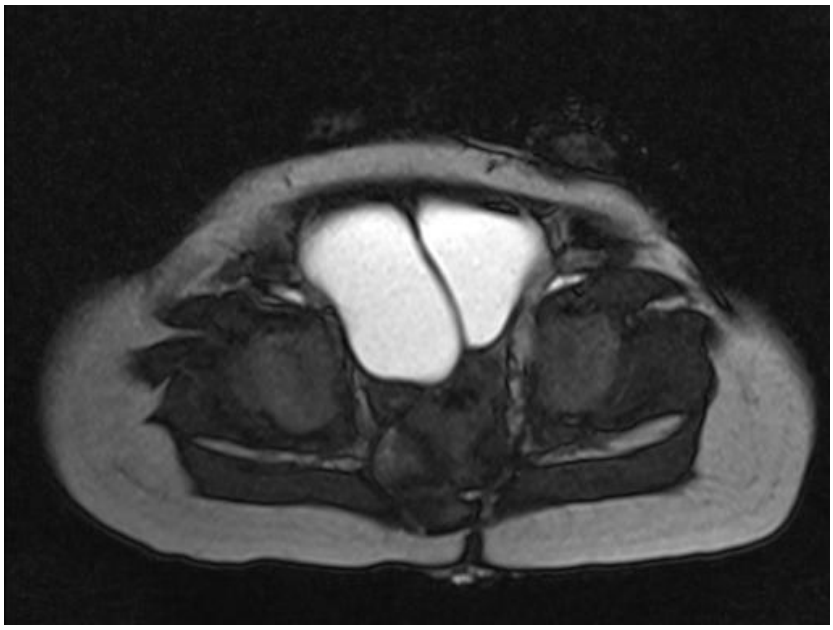


Fig. 3: MRI images of axial sections of T2W sequence, evidence of bladder duplication and uterus didelphys

In vaginoscopy, there were two separate vaginas and each hemivagina had a separate cervix. In cystoscopy and ureteroscopy, each urethra had a separate bladder and a distinct ureteral hole. Moreover, in this study, two different rectovestibular fistulas were seen. The rectovestibular fistula was clean and the left side contained fecal matter.

For anorectoplasty, the patient was again subject to surgery in order to remove the fistula.

In the patient's follow-up, there was still fecal discharge in the vestibular area. Barium enema was applied and there was no evidence of a duplicate segment in the colon. Considering clinical signs and suspected diagnosis of duplication that is expected (although rarely), transverse laparotomy was performed at the patient's previous colostomy site. At the terminal ileum site, there were two separate columns with distinct mesa connected above the peritoneal reflection that had a common proximal opening. One colon had been subject to colostomy in a previous surgery site; therefore, the second colon was resected from ileocecal to the rectum.

3- DISCUSSION

Hindgut duplication is a rare congenital disorder often classified as caudal duplication syndrome with neural tube and anorectal malformation. Caudal duplication is an inclusive term that encompasses duplication of cloaca, urogenital duplication and hindgut. Lower abdominal wall and pelvic bone abnormalities are relatively common (4).

The embryological basis of caudal duplication syndrome is not known. Some researchers believe that this syndrome is caused by incomplete division of monozygotic twins. Pang et al. proposed a unified theory for double spinal cord malformations, all of which result from abnormal linkage between ectoderm and endoderm (5).

Most patients have moderate to severe neurological defects while some of them (like our case) may be neurologically normal (6).

In mild cases of urogenital duplication, isolated urethral duplication occurs almost exclusively in boys. In contrast, urethral duplication in girls is nearly always associated with bladder duplication. Simultaneous skeletal or vertebral anomalies have also been reported in patients with bladder duplication (7). Complete duplication of the colon, rectum, urethra, bladder, uterus, and vagina is highly uncommon in caudal duplication syndrome (8). Anorectal involvement may include anal canal stenosis, ectopic anus, or imperforated anus.

Two types of colon duplication have been reported, including partial or complete involvement of the colon and rectum (type 1) and the forms associated with genitourinary duplication (type 2). In addition to genitourinary abnormalities, spinal malformations, omphalocele, separation of symphysis pubis and VACTERL association have been reported along with colon duplication in 80% of cases.

The tubular type is the most common form of colon duplication and is more prevalent in females than in males (2:1 ratio) (9). Tubular duplication can be isolated without connection with adjacent intestinal segments or in relation with the main colon and connected with one or more openings. Fistula connections with other systems are rare but should be considered (10).

In 1960, Bryndorf and colleagues reported a case that had two anuses in the perineum and several reports have since been made on this subject (1).

In 1986, Yucesan et al. reported the case of a one-year-old girl with complete tubular duplication of the colon. The patient was referred due to excretion problems and a fistula to anus was visible upon

examination. In 2004, Kaur et al. reported the case of a three-month-old baby girl with excretion problems in whom excretion to perineum was observed through two openings. In a 2005 study by Sarpel, a baby girl was reported with complete duplication of colon, with one end opening in the rectum and the other ending in vestibular fistula. The two colons had a common wall but were not connected to each other. In our case, however, the two colons were separated from each other and did not have a common wall (11).

In most patients of this group, spinal malformation is expected; nevertheless, there was no cord or spinal deformity in our case.

Patients' clinical symptoms are variable and depend on the type of colon duplication. Patients with >2 years of age may present with symptoms of constipation, vomiting, abdominal pain, bleeding, lulus, perforation, or obstruction.

Imaging plays a crucial role in the detection of this malformation, which is not always easy to diagnose. Barium enema is often used to evaluate the colon (12).

Despite normal barium enema results, the diagnosis of complete colon duplication was considered for our patient according to her clinical findings, which is a very rare anomaly and is interesting in this regard. Another noteworthy point is the presence of fistulas to other organs in this syndrome that should always be taken into account. In our case, there were two separate rectovestibular fistulas, which resulted in persistent fecal discharge despite colostomy treatment.

4- CONCLUSION

Colon duplication rarely occurs and is usually associated with genitourinary duplication and complex congenital anomalies. It also poses a diagnostic and therapeutic challenge for the pediatric surgeon and may not be easy to diagnose. Therefore, the wide range of anomalies must always be considered. In most cases, numerous reconstructive surgeries are performed to eliminate complications for improving the appearance as well as for cosmetic reasons.

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