

Case Report

Caudal duplication syndrome: imaging evaluation of a rare entity in an adult patient

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**ABSTRACT**

Several theories have been put forth to explain the complex yet symmetrical malformations and the myriad of clinical presentations of caudal duplication syndrome. Hereby, reported case is a 28-year-old female, gravida 2 para 2, with congenital caudal malformation who has undergone partial reconstructive surgeries in infancy to connect her 2 colons. She presented with recurrent left lower abdominal pain associated with nausea, vomiting, and subsequent feculent anal discharge. Imaging reveals duplication of the urinary bladder, urethra, and colon with with cloacal malformations and fistulae from the left-sided cloaca, uterus didelphys with separate cervices and vaginal canals, right-sided aortic arch and descending thoracic aorta, and dysraphic midline sacrococcygeal defect. Hydronephrosis of the left kidney with left hydroureter and inflammation of one of the colons were suspected to be the cause of the patient’s acute complaints. She improved symptomatically over the course of her hospitalization stay with conservative treatments. The management for this syndrome is individualized and may include surgical intervention to fuse or excise the duplicated organs.

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**Introduction**

Caudal duplication syndrome (CDS) is a rare disease entity with prevalence of less than 1 per 100,000 at birth. It encompasses a wide spectrum of anomalies and clinical manifestations, from partial or isolated duplication of organs within the gastrointestinal (GI), genitourinary (GU), and distal neural tube system, to complete duplication. Those anomalous structures can usually be demonstrated by imaging modalities including transabdominal or transvaginal ultrasound, computed tomographic scan, and/or magnetic resonance imaging (MRI). 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 process that disrupts the embryogenesis. In most cases, reconstructive surgeries are performed to address the common complications associated with CDS including imperforate anus, fistula, and obstruction within the GI and/or GU tracts, to improve the cosmetic appearance...
in the case of genital duplication, or to help correct infertility associated with anatomic anomalies.

**Case report**

The patient is a 28-year-old female gravida 2 para 2 with medical history of congenital (GI) and GU system malformation who presented with acute onset of left lower quadrant pain, nausea, and vomiting for the past several days. She endorsed 3 similar episodes in the past 10 years, some of which were associated with urinary tract infections (UTI). After being examined, she started to develop continuous feculent discharge from her anus which was accompanied by some relief of her presenting pain. Surgical history is significant for serial anal dilatation for anal stenosis at birth, 2 reconstructive surgeries for anastomozing her 2 colons, and 2 caesarean sections with classical incisions.

On physical examination, her abdomen was soft with diffuse rebound tenderness but no guarding. Hyperactive bowel sounds were present diffusely across the abdomen. The patient had 2 vaginal openings with labia lateral to each vagina and a third labial fold in between. Two gluteal clefts with a fat pad in between the clefts were noted. One anal orifice was present in the right gluteal cleft; however, the anal sphincter was not appreciated on internal and external palpation. There was no perforation in the left gluteal cleft. During colonoscopy, evidence of prior surgery was seen with 2 orifices noted immediately after intubating the anus. One of them was unable to be cannulated, whereas the second one was strictured, friable, and erythematous. Fecalith of approximately 3 cm in the rectum and diffuse areas of ulcerated mucosa with pseudomembranes, particularly in the distal colon, were noted.

The patient has carried 2 pregnancies in the right uterus, and she normally uses the right vagina for intercourse. She passes urine from both the left and right vagina, although the urinary stream on the right is stronger. Prior episodes of UTI are believed to have occurred from the left urinary system. She is unable to state how she normally passes stool.

**Investigations and/or imaging findings**

A pelvic ultrasound was performed, followed by contrasted computed tomographic scan of the abdomen and pelvis and the pelvic MRI with and without contrast. Mullerian anomalies were demonstrated significant for uterus didelphys with 2 cervices (Figs. 1 and 2). Two urinary bladders were visualized, the left one with a trabeculated wall (Fig. 3). The left kidney demonstrated mild hydronephrosis with hydroureter which drained into the left bladder (Fig. 4A). A small nonenhancing mass just inferior to the right kidney that resembled a tiny dysplastic kidney had a subtle atrophic vascular connection to the inferior vena cava (Fig. 4B). The complete colon was also duplicated, with one colon containing normal well-formed stool. The other colon appeared distended with fluid, had wall thickening with mild surrounding inflammation, and contained a fecalith in the rectum (Fig. 4). Midline fusion defects of L5 and the sacrum were also noted (Fig. 5). The descending aorta was right sided at the diaphragmatic hiatus, and chest radiograph demonstrated a right-sided aortic arch (Fig. 6). MRI of the pelvis demonstrated cloacal malformations with fistula from the left-sided anus to the left cloaca (Fig. 7A) and additional fistula from the left cloaca to the external skin surface (Fig. 7B).

**Treatment**

The patient was admitted for inpatient monitoring and stabilization. Intravenous ciprofloxacin and metronidazole were administered for colitis. Her abdominal pain and anal discharge gradually resolved with the removal of the fecalith during colonoscopy and with the drainage of the colonic fluid. Over the next couple of days, she resumed normal bowel movements with formed stool. Random biopsies of the colon showed active colitis, most likely a consequence of stasis and bacterial overgrowth in the duplicated colon. The patient was provided with the recommendation from the Pediatric and Colorectal Surgery teams to have surgical intervention with removal of the duplicate colon; however, she preferred to be discharged from the hospital and be followed up as an outpatient.

**Fig. 1** (A) Transverse transabdominal sonographic image shows duplicated uterus and bladder (arrows). (B) Transverse transvaginal sonographic image shows duplication of the cervix (arrows).
Discussion

This female patient demonstrated a case of duplicated organs involving the GI and GU systems and anomalies of the vertebral column with preserved fertility. The constellation of findings is considered to be the consequence of abnormal embryogenesis [1]. The embryonic cloaca develops during the third-fourth week of gestation and forms a common channel that gives rise to the GU and GI tracts from the anterior and posterior portions, respectively, as the urorectal septum migrates caudally [2]. The common embryologic origin explains the frequent association of anomalies involving these systems. It has been reported that bladder duplication coexisted with hindgut duplication in 40-50% of the cases [3]. The first case of such organ duplication was reported in “Ephemerides” of Leopoldine Academy at Frankfurt in 1712, and later case reports included a wide spectrum of presentation involving partial to complete duplication of the GI, GU, and distal neural tube systems that Dominguez et al. [4] termed CDS in 1993. CDS is a rare anomaly that encompasses various extent of duplication of the structures derived from the embryonic cloaca and notochord. Its prevalence at birth is less than 1 per 100,000, and to date only about 40 cases have been reported in the literature with slight female predominance in a ratio of approximately 2:1 with no familial and racial predilection, nor other consistent risk factors [1,5–8].

In the least severe form of urogenital duplication, isolated-urethral duplication has been reported that occurs almost exclusively in boys. In contrast, urethral duplication in girls is almost always associated with bladder duplications. Concomitant vertebral or other skeletal anomalies have also been noted in patients with bladder duplication [9]. The complete duplication of the colon, bladder, urethra, uterus, and vagina in CDS is rather unusual [4,10,11]. The acute clinical features of this case presentation are typical of those associated with GU defects found in CDS, including UTI or obstruction, incontinence, and other voiding disorders. Presentations involving the anorectal system may consist of anal stenosis, ectopic anus, and other variants of imperforate anus [11]. This case is unusual in that it offers a unique perspective of CDS patients who do not undergo full reconstructive surgery. A survey of literature found that most reported cases were brought in for treatment in infancy or early childhood, and only 4 cases reported CDS in adults, including 1 patient who had the cloacal malformation repaired as an infant [7,12,13].

The mechanism of the disturbance in embryogenesis leading to considerable anatomic variations is yet to be elucidated, but several etiologies have been proposed. One of the commonly accepted explanations is an incomplete separation of mono-ovular twins or conjoined twinning as a result of misexpression of one or more of the distal HOX genes, potentially HOX10 and HOX11 [5,7,8,12]. Dominguez et al. proposed that an insult to the urorectal septum occurred at approximately 23rd-25th day of gestation [4,8]. Others have suspected that the partial or complete duplication of the

![Coronal T2-weighted fast spin echo MR demonstrates uterus didelphys (arrows) and duplicated bladder (dashed arrows). The left bladder appears trabeculated.](image1)

![Axial contrasted computed tomographic image with (A) left hydronephrosis (B) tiny dysplastic kidney on the right side (arrow).](image2)
organizing center within the embryonic disc may increase the risk of mesodermal insufficiency, and thus account for the failure of complete development of the cloacal membrane and consequent exstrophy [1]. Imperforate anus with fistula into the urinary tract are thought to be the result of abnormal regression of the transient gap (neurenteric canal) during the formation of mesoderm and the shift of the dorsal cloacal membrane [14]. Another causative factor might be the abnormal adherence between the ectoderm and endoderm that resulted in the spinal cord duplication disorders, as suggested by Pang et al [8]. Bremer has postulated that under normal circumstances, portions of the intestinal tract become obliterated at 6th-7th week when vacuoles appear in these cell masses and subsequently coalesce to reconstitute a single lumen. However, if one or more of the vesicle becomes pinched off, a second lumen is created [7,15].

Treatment of CDS consists of patient-tailored, multistage correction of duplication anomalies toward an anatomic, functional, and esthetic reconstruction [6,11]. The urinary bladder septum is removed to create a single chamber. The duplicated colons are usually not fused and have separate blood supply, and either stripping of mucosa or resection of

Fig. 4 – (A) Axial contrasted computed tomographic image demonstrates duplication of the rectum (arrows) with fecalith in the left rectum. Midline sacrococcygeal defect is also noted. (B) Coronal reconstructed computed tomographic image demonstrates duplication of the bladder (dotted arrows) and colon with the normal appearing stool-filled colon (solid arrows) and the dilated, fluid-filled colon (dashed arrows).

Fig. 5 – Three-dimensional reconstruction of pelvic computed tomographic illustrates the midline fusion defects of the L5 vertebral body and the sacrum.

Fig. 6 – Chest radiograph shows right-sided aortic arch (arrow) and descending aorta.
duplicated colon and rectum can be undertaken [15]. If left untreated, some common complications associated with duplicated hindgut include constipation, obstruction, volvulus, and neoplastic changes [10]. On the other hand, the purpose of the reconstructive surgery for genital duplication is mainly cosmetic, as normal menstruation, coitus, and pregnancy are anticipated in most female patients of CDS, [12] although infertility work-up has also been reported [7]. Although our patient had undergone caesarean section, Greenberg et al. [13] demonstrated that patients are capable of successful vaginal delivery after surgical repair of cloacal malformation. Similar surgery can be performed in male patients to remove one of the hemiphalluses for cosmetic reasons [7,15]. Although an overall good surgical outcome is reported, the controversy in treatment is mainly centered around whether the risks and burdens of potential complications of surgery outweigh the psychological impact on the patient’s self-esteem and quality of life [6]. In this case, our patient has declined further reconstructive surgeries at this time as she considers her current condition satisfactory.

REFERENCES


Fig. 7 – (A) Axial T1-weighted postcontrast fat suppressed spoiled gradient echo MR demonstrates fistula from the cloaca to the anus (arrow). (B) Axial T2-weighted FS fast spin echo MR demonstrates cloacal-cutaneous fistula (arrow).