

Caudal duplication syndrome: A rare entity

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Abstract

The rare caudal duplication syndrome is a spectrum of anomalies primarily involving partial or complete duplication of organs comprising the gastrointestinal, genitourinary and distal neural tube systems. These findings are considered to be a result of aberrant embryogenesis. We hereby report a case of an adult female with complete duplication of the genital and urinary systems (urethra and bladder), hindgut and lower end of vertebral column with no functional impairment. She presented in her first pregnancy at 36 weeks gestation, in labour. To the author's knowledge this is the first case of caudal duplication syndrome with pregnancy from Pakistan.

Keywords: Caudal duplication, Genitourinary, Gestation, Hindgut.

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Introduction

The rare caudal duplication syndrome (CDS) is an association between malformations and duplication of gastrointestinal, genitourinary systems and neural tube defects.¹ Reported prevalence at birth is less than 1 per 100,000 but the condition seems to be more rare.² Female to male ratio is approximately 2:1 with no familial or racial predisposition and no other concordant risk factors.³ CDS is proposed to occur as a result of foetal insult at approximately the 23rd-25th days of gestation. It is interesting that the duplicated organs at the site of origin are initially separated by a septum and later branch as independent organs.⁴ During hindgut formation, the cloaca divides into the urogenital sinus and the anorectal canal from posterior and anterior portions, respectively.⁴ The common embryologic origin explains the frequent association of anomalies involving these systems.⁵ Genetic disorders and conjoined twinning have been proposed in the aetiology, in addition to other factors.⁶ Since 1953, the total number of reported cases is less than 100 with varied clinical presentations.⁷ Most of these cases are diagnosed

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in paediatric age groups. We report the case of a female patient with CDS who presented at 36 weeks of gestation, in labour. To our knowledge, cases of asymptomatic presentation of CDS in adulthood are very rare.

Case Report

Thirty-one years old, non-booked primigravida reported to Federal government polyclinic hospital Islamabad in the emergency department on 18 August 2022 where she subsequently received medical care. She had been married for one and a half years and now presented at 36 weeks of gestation with pre labour rupture of membranes for the last 12 hours. She was vitally stable with symphysiofundal height of 36 cm, and longitudinal breech. Foetal cardiac activity was positive with cardiotocograph (CTG) reactive. Her local examination revealed duplication of vulvar, urethral and anal orifices (Figure-1). As she was draining clear liquor from left introital orifice, bishop scoring was done first on this side (4/12) with absent membranes while on the right side there were no signs of labour.

Her ultrasound showed a breech baby with reduced liquor in the left sided uterus.

Right sided uterus was of normal size with thickened endometrium. All baseline investigations were normal. She was catheterized preoperatively separately from both urethral orifices with drainage of urine from each side. With a diagnosis of primigravida breech in labour, an emergency caesarean section was performed. Per operatively a didelphys uterus (Figure-2) and two bladders were seen, with one of the bladders located at abnormally high level.



Figure-1: Duplicated external genitalia localised laterally and each having a urethral, vaginal and anal orifice. There is a wide gap between pubic bones (confirmed by CT films) as shown by the space between the two genitalia.

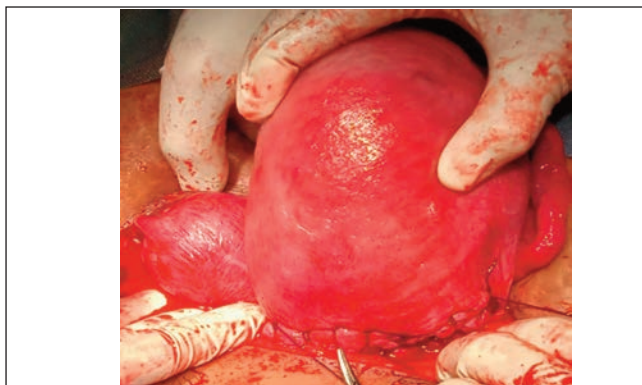


Figure-2: Left side: Small non-gravid uterus, Right side: Enlarged gravid uterus after surgery.



Figure-3: Axial computed tomographic image demonstrates complete duplication of urinary bladder, uterus and rectum.

She delivered a baby girl as breech with a good APGAR (Appearance, Pulse, Grimace, Activity and Respiration) score and normal looking external genitalia. During the post-operative period, her ultrasound and computed tomography (CT) scan (abdomen and pelvis) were performed on the fourth post-operative day for further evaluation. Computed tomograph (CT) scans revealed complete duplication of the uterus, cervix, vagina, rectum and anal canal. Ureters of both sides drained into ipsilateral bladders which opened to the exterior through a urethra on each side. Malrotated bilateral kidneys along with dysmorphic distal sacral vertebrae and complete duplication of coccyx was seen.

Barium studies revealed complete duplication of hindgut (from anus to caecum) with two ileocaecal junctions, one in the left half and other in the right half of abdomen. The last part of the terminal ileum was also duplicated.

Past history: Her life so far had been uneventful. Since her birth she had two separate but concurrent streams of urine without any difference. She passes stool from two anal openings simultaneously or at different times but in the same sitting. There was no functional neurological

impairment. As she belonged to a low socioeconomic class, her abnormal findings were hidden to prevent social stigma. Her family history was unremarkable. Her puerperium was uneventful and now she is on a follow-up programme, living a healthy life with her family.

Discussion

This patient demonstrated duplication of organs involving gastrointestinal, genitourinary system and anomalies of vertebral column without any functional impairment and with preserved fertility. In 40-50% of cases of CDS, bladder and hindgut duplication co-existed.⁵ A classic case of CDS exhibits malformations of the spinal column due to damaged notochord in addition to the duplication of structures which originate from primitive intestine.¹ The complete duplication of colon, bladder, urethra, vagina, cervix and uterus in CDS (as in our case) is rather unusual.⁵

The duplication of colon can be cystic or tubular which is mostly seen on mesenteric side with common vascular supply.⁸ Surgical management for the duplicated colon which is not fused and has a separate blood supply, can be achieved by stripping the mucosa or by functional emptying by stapling or removing the duplicated segment.⁹

In a systematic review of 23 cases, hindgut duplication always involved the anorectal region. Colon duplication extended from the anal region to the transverse colon or ascending colon in most cases and less to the terminal ileum but in the index case, there was complete duplication of hindgut extending from anus to terminal ileum. In females, genital duplication was present in all cases. In all adult females the reason for presentation was related to pregnancy (complications after a successful vaginal delivery, fertility evaluation) or late complications (obstruction of colon anastomosis or repeated UTI with renal scarring).¹⁰ Spinal cord malformations were reported in half of the cases.¹⁰ The level of duplication in the spine is mostly in the lower thoracic region, similar to the branching of a tree.⁴ Defects of fusion and hemivertebrae of vertebral spine were most common, like in our case where the 3rd sacral vertebra was dysmorphic along with coccyx duplication.

The management of each patient with duplication anomalies is individualised. The purpose of treatment is to preserve or improve faecal and urinary continence, maintain reproductive potential, allow a satisfactory sexual life with acceptable cosmetic appearance of the perineum and manage other neurological disabilities.¹⁰ In the index case no surgical intervention for anomalies correction was done as she was asymptomatic.

The purpose of reconstructive surgery for genital duplication is cosmetic as normal menstruation and pregnancy are anticipated in most female patients of CDS although infertility workup has also been reported. Although our patient has undergone caesarean section due to malpresentation, Green et al demonstrated that patients are capable of successful vaginal delivery after cloacal membrane repair.¹¹

Conclusion

Complex malformations affecting multiple caudal organs may have a strong impact in many aspects of the long-term quality of life but as the condition is rare there is paucity of data. Despite multiple anomalies involving different organ systems, patients with CDS have potential to conceive spontaneously, achieve full term pregnancy, deliver without complications and without the need for reconstructive surgery especially in asymptomatic cases which are quite rare. This young patient with asymptomatic presentation of CDS was unique as well as rare. She conceived spontaneously, achieved near term pregnancy and delivered by caesarean section due to malpresentation. She did not require any reconstructive surgery related to any of the organ systems involved as she was asymptomatic. However, such patients require multidisciplinary approach planned ahead of any intervention. Therefore, there is a need for increased awareness among health professionals regarding diagnosis of CDS (from paediatric to adult age range) and management of these cases with a multidisciplinary approach.

Consent: Consent was taken from the patient to share sensitive photographs.

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Conflict of Interest: None.

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