Complete urogenital and colonic duplication: An extremely rare developmental anomaly in an adult female

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ABSTRACT

Complete urogenital duplication is an extremely rare congenital syndrome, where the etiology is unambiguously not explained. Misexpression of certain genes and teratogenic factors are presumed to have a prominent effect on duplications and malformations at various stages during embryogenesis. Several cases have been reported on gastrointestinal and genitourinary duplication. However, no two cases described in literature are alike and they differ with the level of duplication and the associated anomalies. Interestingly, most of the cases reported are at infancy and adult caudal duplication syndrome (CDS) cases are exceptional. Herein, we report a case of a 36-year-old unmarried female with duplication of external genitalia associated with duplication of the genitourinary and colonic system. Imaging revealed associated skeletal and spinal anomalies. Patient did not have any reconstruction surgeries previously and consulted us regarding the possibility of intercourse and fertility. The extent of urogenital duplication and associated anomalies was delineated with help of X-ray, abdominal and trans-perennial ultrasound and magnetic resonance imaging (MRI).
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Complete urogenital duplication is an extremely rare congenital syndrome, where the etiology is unambiguously not explained. Misexpression of certain genes and teratogenic factors are presumed to have a prominent effect on duplications and malformations at various stages during embryogenesis. Several cases have been reported on gastrointestinal and genitourinary duplication. However, no two cases described in literature are alike and they differ with the level of duplication and the associated anomalies. Interestingly, most of the cases reported are at infancy and adult caudal duplication syndrome (CDS) cases are exceptional. Herein, we report a case of a 36-year-old unmarried female with duplication of external genitalia associated with duplication of the genitourinary and colonic system. Imaging revealed associated skeletal and spinal anomalies. Patient did not have any reconstruction surgeries previously and consulted us regarding the possibility of intercourse and fertility. The extent of urogenital duplication and associated anomalies was delineated with help of X-ray, abdominal and trans-perennial ultrasound and magnetic resonance imaging (MRI).

Keywords: Caudal duplication syndrome (CDS), Developmental anomalies, Duplication of external genitalia, Genitourinary and colonic duplication

INTRODUCTION

Caudal duplication syndrome (CDS) is a very rare congenital abnormality that is associated with duplication and malformation of caudal structures comprising of the spine, the spinal cord, gastrointestinal and urogenital systems [1]. The etiology of CDS is not completely understood. However, it is assumed that misexpression of HOX genes encoding for the transcription factors that regulate the developmental process may have a prominent role in it [2]. The prevalence of this syndrome is less than 1 in 100,000 births and adulthood CDS reports are extremely rare. In this article, we present an adult female with caudal duplication syndrome.
CASE REPORT

A 36-year old unmarried woman approached our clinic for evaluation of external genital malformation and possibility of intercourse in the future. A detailed physical examination revealed a double vulva with a 15-cm intervening area comprising of a pad of fat covered by hairless skin. Each vulva had pubic hair, a separate clitoris, urethra, vagina and anal orifice (Figure 1). The patient gave a history of passing two separate streams of urine during micturition, dual passage of feces from both anal orifices and menstruating through both the vaginas. She had an absent pubic symphysis and a non-surgical irregular scar on her lower abdomen (Figure 1) and there was no umbilicus.

Trans-abdominal pelvic sonography showed the presence of two separate bladders, two uteri and multiple follicles in both the ovaries. Two kidneys and two ureters along with colonic duplication with two separate rectums were observed.

Magnetic resonance imaging (MRI) scan (Wipro GE OPTIMA 360 advance 1.5 TESLA) results demonstrated normal kidneys, liver, gallbladder, biliary tract, pancreas, spleen, aorta, inferior vena cava, adrenals, mesentery and omentum. A single esophagus, stomach, duodenum and proximal small bowel with evidence of congenital duplication of the colon from the ileocecal junction ending in 2 separate ani were observed in MRI scan. It also showed duplicated bladders, vagina, uteri with double cervix and ovaries (Figure 2). Fibroids were noted in both the uteri, of which the one in the left uterus is larger (Figure 3). The distal sacrum showed partial agenesis. Ascites or abdominal lymphadenopathy was not observed. We also observed a clear tethering of the spinal cord with low insertion, dural ectasia in the lumbosacral spine and a split dural sac at sacral level and segmentation abnormalities of the lower dorsal vertebra.

Normal levels of FSH, TSH and prolactin were noted during the blood workup. Renal function test was normal.

DISCUSSION

Caudal duplication syndrome typically consists of a combination of several rare malformations and duplications of the distal organs derived from the hindgut, neural tube and caudal mesoderm. Although the specific reasons for caudal duplication are not well-known, it is presumed that an incomplete separation of monovular twins would be the one of the reasons. It is also believed that the impact of genetic, environmental and teratogenic factors influence the duplication of the embryonic cloaca and notochord at various levels during embryogenesis.

Even though caudal duplication syndrome is a rare condition, majority of the cases reported are soon after the birth. Reports on asymptomatic urogenital duplication in adulthood are uncommon. Early detection of malformation and duplication of organs may increase
the chances of successful corrections, especially in cases of urogeneital duplications. A study by Greenberg et al. (1997) [3] showed successful vaginal delivery of a patient where the cloacal malformation was repaired at infant stage. Very few cases of complete urogenital duplication in an adult are reported all around the world till date [4]. In this report, we presented a 36-year-old unmarried female with non-symptomatic complete external and internal duplication of the urogenital system with fibroids developing in both the uterus. We delineated the extent of the colonic, genitourinary and skeletal anomalies by using multimodal imaging. Duplication of the external genitalia in this particular case is highly unique with internal urogenital doubling and skeletal anomalies. While reconstructive genitourinary surgeries are performed to prevent complication and address the fertility. In this particular case, although the reproductive physiology, menstruation and gonadotropin levels are normal, due to the abnormal urogenital anatomy it is difficult to anticipate normal coitus. However, there has been case report published of successful cesarean delivery of an adult woman with duplication of the urogenital system [5, 6]. As the fibroids are asymptomatic patient was not advised any treatment. Since both the uterus has fibroids, this case raises the interesting possibilities of an association of the development of fibroids in anomalous uterus.

**CONCLUSION**

It is beneficial to diagnose of malformations and duplications of caudal structures in infancy to allow possible early surgical correction. Improved non-invasive imaging as seen here has opened an era of new possibilities for diagnosing, delineating and monitoring such complex anomalies both in childhood and adulthood for better treatment strategies in the future.

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Aarthi Deenadayal Tolani – Substantial contributions to conception and design, Acquisition of data, Analysis and interpretation of data, Revising it critically for important intellectual content, Final approval of the version to be published

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Vijay Bhasker – Substantial contributions to conception and design, Acquisition of data, Analysis and interpretation of data, Revising it critically for important intellectual content, Final approval of the version to be published

**Guarantor**

The corresponding author is the guarantor of submission.

**Conflict of Interest**

Authors declare no conflict of interest.

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