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A newborn with caudal duplication and duplex imperforate anus

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Abstract There are case reports of duplication of the colon, rectum, anus, urinary system, lower genital tract, and external genitalia, spinal anomalies, and abdominal wall defects. However, it is rare to encounter a single newborn with all of the mentioned abnormalities, which have been defined as the caudal duplication syndrome (CDS). Herein, we present a newborn with an omphalocele, duplex external genitalia (with duplex labia minora and labia majora), duplex urethral orifices, duplex vaginal orifices, and duplex anal dimple with imperforate anus and rectovestibular fistula on both sides. Exploration revealed duplex appendix, colon duplication, duplex uterus (continuing with tuba and ovaries on both sides), duplex rectum, malrotation of the intestines, with the cecum located in the middle of the abdomen, defect in the intestinal mesentery, and internal herniation of the small intestines through this defect. The intestines were operatively reduced and the defect repaired.

There are case reports of duplication of the colon, rectum, anus, urinary system, lower genital tract, and external genitalia, spinal anomalies, and abdominal wall defects. However, it is rare to encounter a single newborn with all of the mentioned abnormalities, which have been defined as the caudal duplication syndrome (CDS) [1]. CDS is a rare entity in which structures derived from the embryological cloaca and notochord are duplicated in varying degrees [2]; in other words, it indicates partial or complete duplication of the spine and/or spinal cord and of other caudal structures like the gastrointestinal and urogenital tracts [3]. Herein, we report a newborn with CDS, and additionally, duplex imperforate anus and duplex rectovulvar fistulas.

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1. Case report

A 3020 g female newborn, born by cesarean section to a 22-year-old mother, was referred to our hospital with an omphalocele and duplication of external genitalia and anorectal region with imperforate anus on both sides. The physical examination revealed an omphalocele 4 cm in diameter, duplex external genitalia (with duplex labia minora and labia majora), duplex urethral orifices, duplex vaginal orifices, and duplex anal dimple with imperforate anus on both sides (Fig. 1). Fecal drainage was observed from the orifices near the left and right vaginas (Fig. 2). Urine outflow was noted from both right and left urethras. Abdominal ultrasound revealed normal kidneys and duplex uterus. Echocardiography was reported as patent ductus and apical ventricular septal defect. As the patient had defecation throughout rectovulvar fistulas on both sides, enteral

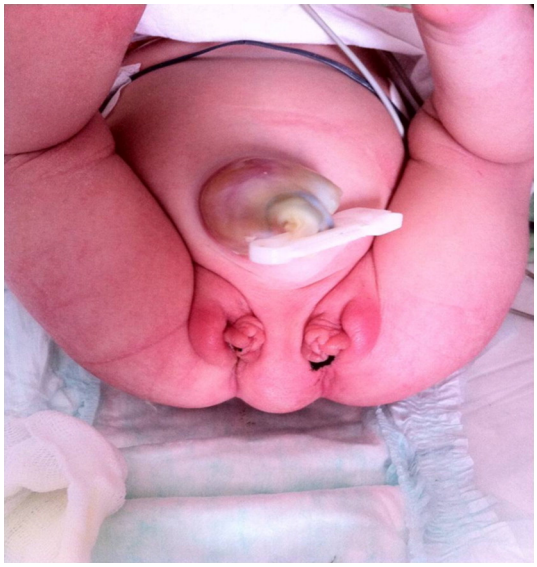

Fig. 1 Physical examination of the patient showed an omphalocele 4 cm in diameter, duplex external genitalia (with duplex labia minora and labia majora), duplex urethral orifices, duplex vaginal orifices, and duplex anal dimple with bilateral imperforate anus. Fecal drainage was observed from the orifice near the left vagina.

feedings were started and were well tolerated. Vertebral radiographs showed vertebral dysplasias and bifid L5 and sacrum (Fig. 3). Cytogenetic studies revealed a normal 46, XX karyotype. Exploratory laparotomy and omphalocele repair were done on the postnatal 3rd day. Before laparotomy, examination under general anesthesia was performed and both left and right urethral and vaginal orifices and rectovulvar fistulas on both sides were catheterized. Both anal regions were examined with electrical stimulation, and it was seen that a duplex muscle

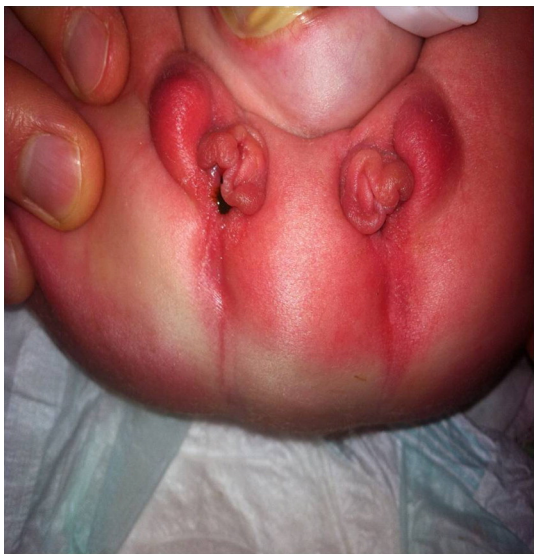

Fig. 2 Physical examination of the patient showed duplex anal dimple with bilateral imperforate anus. Fecal drainage was observed from the orifice near the right vagina.

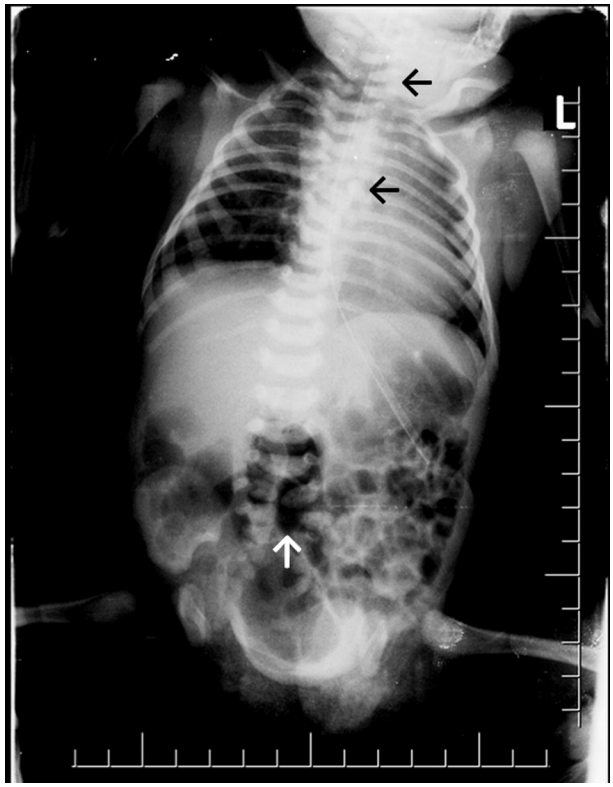

Fig. 3 Vertebral radiographs showed : vertebral dysplasias (black arrows) and bifid L5 and sacrum (white arrow) are seen.

complex was present and reactive on both sides, though the right-sided muscle complex showed a better response. Both right and left rectovulvar fistulas were calibrated with Hegar dilators. A laparotomy was done and the omphalocele excised. The exploration revealed duplex appendices (Fig. 4), sigmoid and cecum duplication (as seen externally) (Fig. 5), duplex uterus (continuing the fallopian tubes and

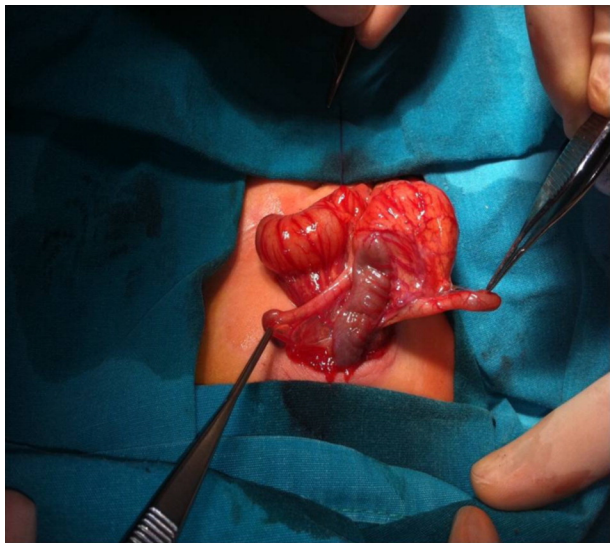

Fig. 4 At exploratory laparotomy a duplex appendix was observed.