

Caudal duplication syndrome

Spinal and spinal cord duplicity ([diastematomyelia](#)) malformations span a wide spectrum of anomalies, ranging from a simple fibrous band splitting the cord into halves to complete duplication of the spine and spinal cord. The more serious forms are rare and only a limited number of cases are on record.

They are usually associated with other systemic malformations, including duplication of vascular structures, of the distal gastrointestinal and urogenital tracts, and possibly limb malformations. The term caudal duplication syndrome has been applied to those instances.

This entity was first described by Dominguez et al. from the Department of Pediatric Radiology, University of [Texas](#) Southwestern Medical Center, [Dallas](#). They reported 6 new cases as well as reviewed 8 already reported cases of multiple anomalies and duplication of the distal organs derived from the hindgut, neural tube and adjacent mesoderm ¹⁾.

Caudal duplication was considered a rare type of [conjoined twins](#) previously ²⁾ in which structures derived from the embryonic cloaca and notochord are duplicated to various extent.

The term encompasses a spectrum and often is quoted as one type of incomplete separation of monovular twins. Bajpai et al., present more evidence giving credence to caudal twining as the mechanism behind the syndrome. ³⁾

Epidemiology

Caudal duplication syndrome is a rare condition with only about 40 cases reported in the literature ⁴⁾.

Female patients predominate in a ratio of about 2:1, and no familial or racial predilection has been shown ⁵⁾.

Etiology

Exact etiology of caudal duplication syndrome is unknown. Various theories have been suggested. Incomplete separation of monozygotic twin has been postulated as the etiologic factor ^{6) 7)}.

Pang et al. advanced a unified theory for the spinal cord duplication disorders, suggesting that all result from abnormal adherence between ectoderm and endoderm ⁸⁾.

In the view of Dominguez et al. these anomalies originate from damage to the mass formed by caudal cells and posterior gut at approximately 25 days of pregnancy ⁹⁾.

Pathogenesis

Pathogenesis is unclear. Polytopic primary developmental field defect or a disruption sequence or

somatic or germ line mutations in certain developmental genes could be involved ¹⁰⁾.

Partial or complete duplication of the organizing centre within a single 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 or other aberrations ¹¹⁾.

Clinical features

It is a range of disease comprising gastrointestinal, genitourinary, vertebral, spinal, and limb abnormalities. Common gastrointestinal anomalies include duplication of the colon and rectum, which may be associated with a variety of other anomalies such as imperforate anus, rectal fistula, ventral hernia, omphalocele, duplication of terminal ileum, double appendices, Meckel's diverticulum, intestinal malrotation, and situs inversus.

Urogenital anomalies comprise duplication of the external and internal genitalia, ureters, and bladder and anomalies of the kidney.

Spinal anomalies include [hemivertebra](#), [sacral agenesis](#), [myelomeningocele](#), [diplomylelia](#), duplication of lumbar spine, [butterfly vertebra](#), and [spina bifida](#) ¹²⁾.

Most patients have associate moderate to severe neurodeficit; although some can be neurologically normal ¹³⁾.

Treatment

Treatment consists of staged correction of duplication anomalies. Either stripping of mucosa or resection of duplicated colon and rectum is undertaken. Division of septum in the UB is done to make it a single chamber. Corrective surgery in the form of fusion is done for the external and internal genitalia. Any spinal anomaly is also corrected suitably. Many authors have reported near-normal cosmetic and functional result for these complicated anomalies.

Shah and Joshi ¹⁴⁾ suggested removal of one of the hemi-phalluses for cosmetic reasons in males. No corrective surgery was, however, done in the adult female patient reported by them. Liu et al. reported the case of caudal duplication syndrome in which they did multiple stage correction. The bladder septum was removed, the two hemi-phalluses were fused to form one phallus, and the duplicated colon was excised. The patient also had hydronephrotic left kidney and left megaureter, which were removed. Ultimate outcome is good ¹⁵⁾.

These patients are difficult to manage posing numerous surgical as well as medical management challenges. Child's organ systems are usually working normally. Certain questions arise, should one intervene? When should one intervene? And what should be the best intervention, fusion or excision of accessory organ? Organ duplication syndromes are difficult scenarios to manage. Treatment should always be individualized according to the extent of duplication and functionality of the organ systems involved. The malformations that are potentially life-threatening should be addressed first ¹⁶⁾.

Case series

In six children with multiple anomalies and duplications of distal organs derived from the hindgut, neural tube, and adjacent mesoderm, spinal anomalies (myelomeningocele in two patients, sacral duplication in three, diplomyelia in two, and hemivertebrae in one) were present in all the patients. Duplications or anomalies of the external genitalia and/or the lower urinary and reproductive structures were also seen in all our patients. Ventral herniation (in one patient), intestinal obstructions (in one patient), and bowel duplications (in two patients) were the most common gastrointestinal abnormalities.

Dominguez et al., believe that the above constellation of abnormalities resulted from an insult to the caudal cell mass and hindgut at approximately the 23rd through the 25th day of gestation. They propose the term caudal duplication syndrome to describe the association between gastrointestinal, genitourinary, and distal neural tube malformations ¹⁷⁾.

Case reports

2016

A full-term male presented with combination of anomalies including anorectal malformation, duplication of the colon and lower urinary tract, split of the lower spine, and [lipomyelomeningocele](#) with [Tethered cord](#).

Samuk et al., report this exceptional case of caudal duplication syndrome with special emphasis on surgical strategy and approach combining all disciplines involved. The purpose of this report is to present the pathology, assessment, and management strategy of this complex case ¹⁸⁾.

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 cervixes 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 ¹⁹⁾.

2014

A 3-month-old male infant had presented with the classical form of the disease i.e., duplication of the gastrointestinal, genitourinary system and vertebral column with anterior abdominal wall hernia and a large lipomeningocele ²⁰⁾.

2013

Sur et al., report the case of a baby presenting on the first day of life with complete duplication of caudal structures below the dorsolumbar level ²¹⁾.

A 3-day-old male neonate presented with features of [anorectal malformation](#) and duplication of the external genitalia. He was subsequently diagnosed with complete duplication of the colon, rectum, bladder, and urethra associated with [spinal lipoma](#) ²²⁾.

2009

A 13-year-old boy was born with duplicated colon-rectum and anus, diphallus, hydronephrosis of left kidney with megaureter, double bladders and urethras, and vertebral abnormalities. Multiple-stage correction was performed to remove the duplicated colon and the mucosa of the duplicated rectum. A new colon was reconstructed. The left kidney and megaureter were excised. The septum in the bladders was removed to convert the double bladders into a single bladder. The double phalluses were fused into a single penis. After these staged procedures, the boy is now living a normal life ²³⁾.

A female infant, born by cesarean delivery (dilation dystocia), was referred at age of 24 hours with a history of "imperforate anus". Physical examination revealed duplicity of the vulvar introit (urine output by the right orifice and feces by the left). She was submitted to the following imaging exams: (1) echodopplercardiogram - interatrial and interventricular communications; (2) ultrasonography - pelvic left kidney; (3) barium enema - one of the perineal orifices had a communication with the rectum; the other communicated with the vaginal dome and the bladder (urogenital sinus); (4) voiding cystourethrogram (VCU) - two urethral orifices communicating with the bladder, bladder diverticulum on the right side and vesico-ureteral reflux (grade II) on the left side.

Investigation of the spine was done with conventional radiographs (XR), computed tomography (CT) and magnetic resonance imaging (MRI), which disclosed complex malformations of the thoracic and lumbosacral spine, with "S" shaped dextroscoliolus. Aortic duplication was also noted. The vertebral bodies of T1, T2, T4 and T10 were widened and split by an anterior median incisure. A T7 hemivertebra was also present. From T11 level there was complete duplication of the vertebral bodies extending down to the S2 level. The remaining sacral and coccygeal vertebrae were absent. Duplicated vertebrae were joined posteriorly by deformed laminae and encompassed an extremely enlarged spinal canal. The spinal cord was duplicated from level T1 downwards. From L1 level, a large lipoma occupied the spinal canal and communicated with the subcutaneous tissues inferiorly ²⁴⁾.

2008

In a 2-year-old female child, a case of abnormal mass in the perineum with undeveloped feet, duplication of colon, external genitalia, and lumbosacral vertebra was reported by D'Costa et al. ²⁵⁾.

2006

Shah and Joshi reported the case of an asymptomatic adult female with duplication of colon, rectum, anus, urinary bladder (UB), urethra, uterus, cervix, vagina, and external genitalia ²⁶⁾.

2004

Bajpai et al., report successful surgical management of a full-term infant with a constellation of anomalies of caudal duplication syndrome ²⁷⁾.

Radiographic, CT, and MR images of a 15-year-old girl who had lower back pain showed asymmetric lumbar spine duplication with spinal cord tethering secondary to a filum lipoma in the sacrum. Despite gross spinal abnormalities, the patient was neurologically intact and has been followed up with conservative treatment ²⁸⁾.

2001

An autopsy case of a full-term infant with incomplete caudal duplication syndrome associated with multiple anomalies.

These anomalies included a duplicated penis; double urinary bladder with an attenuated tunica muscularis; duplication of lower bowel with two ilia, appendices and colons; colonic hypogangliosis and left imperforated anus associated with rectourethral fistula. Other anomalies consisted of sacral meningocele, sacral duplication with hypoplastic left sacrum and pelvic bones, muscle atrophy and hypoplasia of the left lower extremity, abnormal lobation of liver with stomach entrapment, omphalocele, and right atrial isomerism syndrome. The complex pattern of anomalies suggests the possibility that partial caudal duplication might be part of the spectrum of conjoined twinning ²⁹⁾.

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