

Caudal Duplication Syndrome

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ABSTRACT

Complete duplication of genitourinary system, colon and vertebral column is a very rare and complex congenital condition termed as "caudal duplication syndrome" with variable presentations. This term is often quoted as a type of incomplete separation of mono-ovular twins or conjoined twinning. It is associated with other congenital malformations of the genitourinary, gastrointestinal and other organ systems. The hereby reported case, 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.

Key Words: *Caudal duplication syndrome. Genitourinary duplication. Colonic duplication. Bifurcation of spinal cord. Lipomeningocele. Anterior abdominal wall hernia. Talipes equinovarus.*

INTRODUCTION

Caudal duplication syndrome consists of duplication of genitourinary system, gastrointestinal system especially hindgut, vertebral column, spinal cord and lower limbs. It is also associated with other congenital malformations like kyphoscoliosis of spine, vertebral body defects like hemivertebrae and bifid vertebral bodies, tethered spinal cord, lipomeningocele, cord compression and cauda equina syndrome. Dominguez *et al.* proposed the term of "caudal duplication syndrome" to describe the association between gastrointestinal, genitourinary, and distal neural tube malformations.¹ To the authors' knowledge, no case from Pakistan has ever been reported so far.

CASE REPORT

A 3 months old male infant presented with multiple birth defects. The child was having duplication of genitalia separated by two large swellings. One of them was involving the lower abdomen and the other was involving the perineum. The patient was passing urine through both urethrae independently. He had two anal openings through which patient passed stools either simultaneously or at different times. He was the seventh issue of consanguineous parents. There was no history of such congenital malformations in the family. He was breastfed and immunized according to EPI schedule.

On general physical examination, his vital signs were within normal limits. His weight was on 3rd percentile (5 kg), length on 0.1st percentile (55 cm) and occipitofrontal circumference (OFC) on 3rd percentile (37 cm) for the age and gender. There was a large

midline abdominal wall hernia in the hypogastrum which had appeared as a result of the bifurcation of rectus abdominus muscle. The skin overlying this hernial swelling was hypoplastic with shiny texture and prominent veins. Both penile shafts were normal in size. On the right side, there was a single testis in the scrotum and on the left side testis was in the inguinal canal. There was also right sided reducible indirect inguinal hernia. Perineal swelling was cystic covered with normal skin with no local lymphadenopathy (Figure 1). Spinal column was involved with scoliosis and bifurcation of the spine below 2nd lumbar vertebra. Power in the lower limbs was 3/5. There was bilateral talipes equinovarus. Respiratory and cardiovascular system examination was unremarkable.

His baseline investigations were normal, X-ray spine showed scoliosis of the spine, hemivertebrae, bifid vertebral bodies and bifurcation of vertebral column below 2nd lumbar vertebra. Abdominal ultrasound showed ectopic left kidney lying in the pelvis with bilateral mild hydronephroureters, duplication of the bladder and a large anterior abdominal wall hernia. Barium enema showed complete duplication of colon (Figure 2). Intravenous urography showed duplication of the bladder with ureters of both sides draining into ipsilateral bladder which opened to the exterior by urethrae passing through its respective penile shaft on each side (Figure 3). CT scan abdomen and pelvis with vertebral column confirmed the previously described findings. It also showed that perineal mass was arising from the lower segment of the spine and was a lipomeningocele. MRI spine showed a tethered spinal cord and confirmed the lipomeningocele (Figure 4).

A comprehensive treatment plan has been devised by multidisciplinary team to surgically correct the congenital malformations of this patient in Military Hospital and Combined Military Hospital, Rawalpindi. Team will consist of Paediatrician, Paediatric Surgeon, Neurosurgeon, Spinal Surgeon, Urologist and Plastic Surgeon.

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Figure 1: Photograph of the patient showing diphallia, two scrota, two anal openings (black and white arrows), suprapubic abdominal wall hernia, right reducible indirect inguinal hernia and a large lipomeningocele in the perineum.

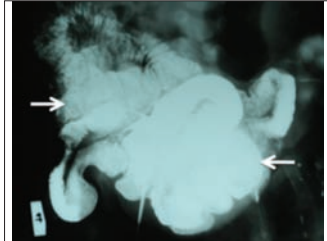


Figure 2: X-ray film of barium enema showing duplication of colon. White arrows showing position of colons.



Figure 3: Intravenous urography film showing ectopic left kidney. Two urinary bladders (white arrows). One can also appreciate bifurcation of the vertebral column.

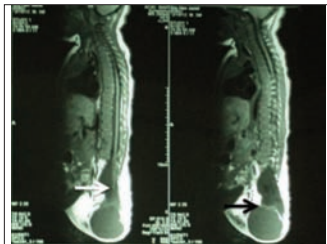


Figure 4: T1 weighted sagittal MRI spine showing anomalous vertebral column, tethered cord (white arrow) and lipomeningocele (black arrow).

DISCUSSION

Caudal duplication syndrome is a rare condition with only about 40 cases reported in the literature.² Dominguez *et al.* 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 Female patients predominate in a ratio of about 2:1, and no familial or racial predilection has been shown.³ Caudal duplication was considered a rare type of conjoined twinning previously,⁴ in which structures derived from the embryonic cloaca and notochord are duplicated to various extent. The term encompasses a spectrum and is often quoted as one type of incomplete separation of mono-ovular twins.² Pathogenesis is unclear. Polytropic primary developmental field defect or a disruption sequence or somatic or germ line mutations in certain developmental genes could be involved.⁵ The insult to the caudal cell mass and hindgut occurs at approximately 23rd – 25th day of gestation.² 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 exostrophy or other aberrations.⁶

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.² They are usually associated with other systemic malformations, including duplication of vascular structures, the distal gastrointestinal and urogenital tracts (as in the present case), and possibly limb malformations. The term caudal duplication syndrome has been applied to those instances.

The embryologic origin of the caudal duplication is not known. Some authors propose that it may result from incomplete division of monozygotic twins. Pang *et al.* advanced a unified theory for the spinal cord duplication disorders, suggesting that all result from abnormal adherence between ectoderm and endoderm.⁷ 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.¹ Pang *et al.* classified spinal cord duplication anomalies into type I and II. The first is characterized by two hemicords, each contained within its own dural sac, and separated by an osteocartilaginous septum. Type-II is defined by two hemicords in the same dural sac, separated by a fibrous septum.^{7,8} The case reported here may be classified as type-I. Female patients are usually infertile or have history of repeated miscarriage. Only one case reported by Ragab *et al.* who attained full term pregnancy.⁹

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.

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