Congenital Pouch Colon with Unilateral Renal Agenesis and Monorchism

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Abstract

Background: Congenital pouch colon, also known as congenital short colon, or "Pouch colon syndrome", is a rare condition that occurs in association with anorectal malformations; colon is either partially or completely replaced by pouch-like dilatation and communicates with the urogenital tract by means of a fistula. This anomaly is exclusively seen in Northern parts of India with only a few cases reported from elsewhere.

Case Presentation: A 1-day old neonate was presented with abdominal distension due to lack of passage of meconium. Clinical and radiological investigations revealed ano-rectal malformation. Incidental findings were left sided renal agenesis and right sided anorchia. Laparotomy revealed congenital pouch colon which was dealt accordingly. The baby is now healthy and awaiting further reconstructive surgery.

Conclusion: Although urogenital anomalies are not uncommon with congenital pouch colon, the finding of renal agenesis with unilateral anorchia is quite rare.

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Key Words: Renal; Agenesis; Congenital; Colonic Pouches; Monorchism

Introduction

Congenital pouch colon (CPC) is a relatively rare form of ano-rectal malformation (ARM) in which part or whole of colon is converted into a pouch-like dilatation that ends blindly and communicates with the urogenital tract by means of a fistula^1,2. Pathogenesis and embryology of CPC are not well understood, but dietary, environmental and familial factors have been hypothesized to contribute to the pathology^2. High number of cases with increased fetal death rate (10-33%) of type IV CPC has been reported^3. Renal agenesis, i.e.,
absence of one or both of the kidneys, is often incidentally diagnosed by abdominal ultrasound or computed tomography (CT) scans secondary to another condition or during routine investigation for other diseases like ARM.

Absence of one or both testes in a phenotypic male with normal karyotype is defined as congenital anorchism[4]. The present study was aimed at reporting a rare case of type IV congenital pouch colon with associated anomaly of unilateral anorchism along with unilateral renal agenesis.

**Case Presentation**

A 1-day old male baby, born of non-consanguineous marriage and from a low economic family, was admitted with history of gradual distention of abdomen, bilious vomiting and failure of passage of meconium with meconuria and pneumaturia since birth. The baby was delivered normally at the 39th week of gestation (BW 2.25 kg; Apgar score 5-7) by a young mother (20 yrs; primigravida, primipara) in a peripheral health center. Prenatal period was uneventful except for lack of proper weight gain. No antenatal ultrasonography was done. On admission, the baby had normal body temperature, high respiratory rates and racing pulse with depressed neonatal reflexes. Physical examination revealed a flat bottom with no anal opening and normal genitalia with empty right scrotal sac. Abdominal examination revealed distended flanks with bulged umbilicus, tympanic on percussion, and sluggish peristaltic sound. The baby also had pneumaturia and meconuria. No family history of congenital abnormalities was reported during admission.

Routine blood count revealed normal hemogram and total cell counts with normal renal function test. The invertogram in AP and lateral view showed a large globular loop of bowel with single air fluid level occupying more than 50% of the total abdominal width. No significant vertebral anomaly was noticed. Chest skiagram revealed normal bony and soft tissue architecture. Abdominal and perineal sonogram showed absence of left kidney and right testis with air in urinary bladder. Intravenous pyelogram, done at 1.6 months age, confirmed absence of left kidney.

Analysis of peripheral blood leukocytes cultured for 72 hrs in presence of mitogen, revealed normal male child with 46,XY karyotype (Fig. 1).

**Differential Diagnosis:**

Anorectal malformation (ARM) with rectovesical or recto-prostatic urethral fistula.

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**Fig. 1:** Karyotype of the subject studied by GTG-banding analysis
Results and Analysis:
Exploratory laparotomy, done on the 2-day old baby, confirmed type IV CPC communicating with bladder neck. Associated malrotation of the midgut without volvulus was detected and left renal fossa was empty. Pouch was excised and low descending colostomy performed, malrotation was corrected (Ladd’s procedure) and appendectomy done. Neither testicular tissue nor structures of spermatic cord was detected in the right posterior abdominal wall.

Follow Up
The baby is presently healthy and undergoing regular monthly follow up. He is awaiting further corrective surgery for anomaly.

Discussion
Congenital pouch colon is an uncommon type of ARM which has been studied in detail in recent times. Higher numbers of CPC cases were reported from India as compared to other countries[2,5-7]. Types I, II and III CPC are usually diagnosed during preoperative evaluation period in ARMs and there are typically large air fluid levels in plain abdominal X-ray[2,8-10].

However, type IV CPC does not have a characteristic appearance on plain X-ray and was diagnosed during surgery for colostomy[3,10].

A few earlier reports have shown the presence of abnormal histology in the excised colon[11]. However, in the present case, excised pouch histopathology revealed presence of normal muscle tissue as well as few ganglion cells.

Incidence of associated cardiac, vertebral and genitourinary anomalies is very high in CPC and therefore, evaluation of these systems is mandatory in all cases of ARM, more so in suspected cases of CPC[2,12]. Unilateral renal agenesis has been reported to be equally common among both sexes[13]. Increased risk of fetal renal agenesis or dysgenesis was observed in mothers addicted to alcohol, smoking etc[14,15]. Lower maternal weight gain during pregnancy has also been reported[14]. In the present case, while no association with addiction was observed, a history of suboptimal maternal weight gain during antenatal period was noticed.

Diagnosis of congenital anorchia is suspected in a patient exhibiting male external genitalia, 46,XY karyotype together with absence of testis in scrotal sac. In addition, a small phallus is a frequent clinical finding in anorchid patients[16].

Surgical exploration performed to repair the CPC in the present case failed to find any testicular elements or Mullerian structures confirming diagnosis of unilateral congenital anorchia on the right side.

Conclusion
Though the incidence of CPC is increasing[3], to the best of our knowledge, presence of type IV CPC with renal agenesis and monorchism in a male boy with normal karyotype is an extremely rare presentation and is described here to bring up to date of the medical fraternity.

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