CONGENITAL POUCH COLON: IS IT REALLY A RARE PATHOLOGY?

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**INTRODUCTION**

Congenital pouch colon (CPC) is a rare condition in which there is a short, pouch-like dilated colon and it is usually associated with an anorectal malformation (ARM) (1-3). The pathogenesis and embryology of congenital pouch colon are not well understood, but dietary, environmental factors and familial inheritance may be contributing factors in this pathology. Most of the cases in the literature have been reported from India. This increased regional incidence may be attributed to the lack of awareness of this pathology or its mislabeling rather than regional distribution. Congenital pouch colon is classified into four types based on the length of the abnormal colon: Type I: Normal colon is absent and the ileum opens directly into the colonic pouch. Type II: The ileum opens into a short segment of cecum which then opens into the colonic pouch. Type III: Presence of a significant amount of normal colon between the ileum and the colonic pouch. Type IV: Presence of nearly normal colon with only terminal portion of the colon (rectum and varying portions of the sigmoid) converted into a pouch. Histopathological examination of CPC reveals mucosal and submucosal inflammation, focal or gene-

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Type IV: Presence of nearly normal colon with only terminal portion of the colon (rectum and varying portions of the sigmoid) converted into a pouch.

Histopathological examination of CPC reveals mucosal and submucosal inflammation, focal or gene-
ralized thinning and disorganization of muscle layer and presence of immature ganglion cells and decreased number of mature ganglion cells with neuronal dysplasia (5). The majority of patients with CPC are reported from India when compared to other parts of the world (1,2,4-7). Although types I and II CPC were more common in large series from India (1,2,4-7), recently the number of patients with type IV CPC has been increasing (8). Hence, is type IV CPC really a rare condition or is it ignored? Here, we report two cases of type IV CPC and discuss the diagnosis and treatment of this condition.

CASE REPORT

Case 1

A one-day-old female presented with rectovestibular fistula. The patient was evaluated for associated anomalies after physical examination. The patient's length and weight were 49 cm and 3.3 kg, respectively. Blood and urine analysis were normal. Plain abdominal X-ray revealed dilated bowel loops. Chest and spinal X-rays, abdominal and spinal ultrasonography (US) and echocardiography were performed. Bilateral hydroureteronephrosis was diagnosed on US examination and bilateral grade-4 vesicoureteral reflux (VUR) was also detected at voiding cystourethrogram (VCUG). Laparotomy was performed through a left lower quadrant oblique incision and type IV CPC was observed. The dilated antimesenteric portion of the sigmoid colon and rectum above the peritoneal reflection level was resected. Tapering coloplasty was completed by closure of the antimesenteric side of the colon over a number 12 anal dilator, and sigmoid diverting colostomy was done. Biopsy specimens revealed mild lymphoid hyperplasia and marked fibrosis in the submucosa. Inflammation with minimal congestion was noted in the submucosa and mucosa. Thinning of the outer muscle layer with a normal inner layer and a decreased number of mature ganglion cells were also noted (Figures 1,2). During posterior sagittal anorectoplasty procedure at the age of six months, the rectum was mobilized from the vestibule, excisional tapering of the rectum below the peritoneal reflection level was completed and the rectum was located within the sphincter mechanism. Colostomy was closed at eight months of age. The postoperative period was uneventful. Barium enema was repeated one year later and the diameter of the rectosigmoid colon was observed to be normal. The child currently has voluntary bowel movements 2-3 times a day and has no soiling.

Case 2

A two-day-old male presented with an anocutaneous fistula ending at the scrotum; orifice of the fistula was quite narrow and was not suitable for dilatation. The patient was examined for possible associated anomalies. The weight and length of the patient were 2.5 kg and 34 cm, respectively. Blood and urine analysis, chest and spinal X-rays and echocardiographic examination of the patient were normal. Abdominal X-ray revealed dilated bowel loops. Left hydroureteronephrosis was diagnosed on US examination and left-sided grade-3 VUR was detected at VCUG. Because of the associated VUR, low birth weight and fistula dilatation problem, we decided to perform a sigmoid colostomy. Laparotomy was performed through a left lower quadrant oblique incision, revealing a greatly dilated rectum and a dilated portion of the sigmoid colon. The dilatation of the sigmoid colon was localized to its lower end and transition to
normal sigmoid colon was abrupt. Type IV CPC was suspected; sigmoid colon biopsy and sigmoid diverting colostomy were performed. Contrast study of the colon after colostomy opening revealed the diagnosis of type IV CPC (Figure 3). Histopathological examination of the biopsy revealed mucosal and submucosal inflammation, thinned outer muscular layer and decreased number of mature ganglion cells. Posterior sagittal anorectoplasty was performed at six months of age and at this stage excisional tapering coloplasty of the rectum and of the distal sigmoid colon was performed over a 13 anal dilator. Colostomy was closed six weeks later. The child has voluntary bowel movements twice a day and no soiling.

**DISCUSSION**

A total of 360 CPC cases have been reported from India, versus only 7 cases from Canada, 6 from Turkey and a few from Iran, China, Japan, United States and Europe (2,9). Types I, II and III CPC are usually diagnosed during the preoperative evaluation period in ARMs and there are typically large air fluid levels in plain abdominal X-ray (1-2,4-7). However, type IV CPC does not have this appearance on plain X-ray and is diagnosed at colostomy opening (7,8,10-12). A variable dilatation of the rectum and sigmoid is always present in ARMs. However, there is no clear definition of a limit for the dilatation of the rectum and sigmoid in ARMs (8). Furthermore, many surgeons do not routinely take a biopsy from a dilated rectum or sigmoid during a colostomy procedure in ARM cases. Many surgeons also prefer a right upper quadrant transverse colostomy instead of a traditional sigmoid diverting colostomy, without exploration of the remaining colon (13). For these reasons, type IV CPC can be easily underdiagnosed. In our cases, we diagnosed type IV CPC based on surgical findings.

Megarectosigmoid is also common after surgical correction in certain types of ARMs - rectovestibular fistula, perineal fistula, rectal atresia, and rectobulbar fistula, and usually occurs if fecal impaction is not prevented and treated by laxatives (13-16). This condition is more common and usually develops after the age of three if patients with ARMs are not followed up appropriately for the earlier recognition and treatment of constipation. In severe forms of this condition, resection of megarectosigmoid is required to reduce the amount of laxatives (13-16). In contrast, type IV CPC is a congenital malformation, is not preventable and surgery is indicated. However, it is still a fact that type IV CPC should be considered in the differential diagnosis of megarectosigmoid in ARMs at any age (7). The discrimination between these two entities again depends on surgical and histopathological findings.

In the histopathological examination of the CPC, the most frequently observed abnormalities are acute and chronic inflammation of the mucosa and submucosa, focal or generalized thinning of muscle layers, especially of the outer muscle coat, disorganized muscle layers, and a decreased number of mature ganglion cells. Neuronal hyperplasia and hypertrophy in nerve plexuses are also observed at the affected segment of the colon (5). We did not perform routine immunohistochemical staining in this report and therefore we could not comment on the presence or absence of neuronal dysplasia. Ho-
wever, we also think that neuronal intestinal dysplasia must be investigated in type IV CPC for correlation with postoperative outcome.

The incidence of associated cardiac, vertebral and genitourinary anomalies is very high in CPC, so evaluation of the genitourinary tract, skeletal system and cardiovascular system for associated anomalies is very important (1,2,5,10). In the systematic evaluation of our patients, both had VUR.

Surgical treatment options in type IV CPC include resection of the affected sites of the colon or excisional tapering coloplasty (2). Reconstruction of the sac-like colon to form a tube to preserve colonic absorptive function is essential (16). For that reason, we prefer excisional tapering coloplasty.

In conclusion, in ARM cases, when the sigmoid colon or rectum is not explored appropriately, associated type IV CPC may be underdiagnosed. However, if type IV CPC is specifically searched, the incidence would perhaps be higher than expected. It is important for gastroenterologists and pediatric clinicians to be aware of the features of this condition to allow for proper diagnostic evaluation and surgical management.

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REFERENCES