Original Article

Congenital Pouch Colon in Girls: An Algorithm for Preoperative Diagnosis

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Abstract

Aim: The aim of the study was o define the preoperative diagnostic clinical and radiological features in girls with congenital pouch colon (CPC).

Materials and Methods: Records of 47 girls with CPC, managed from 1996 to 2018, were reviewed. There were two age groups: Group A (newborn to 12 months; *n* = 26) and Group B (>12 months to 20 years; *n* = 21). The important clinical and radiologic features to help in a preoperative diagnosis were noted.

Results: The most common subtype was Type II (57.4%), followed by Type I (23.4%) and Type III (12.8%). The features common to both the groups were abdominal distension (A = 53.8%; B = 9.52%), severe perineal excoriation (A = 19.2%; B = 23/8%), and urinary incontinence (A = 30.7%; B = 85.7%). In addition, in Group B, fecaloma on abdominal palpation was noted in 28.6% of patients. The characteristic appearance of the perineum including external genitalia and findings on plain abdominal X-ray (AXR) were 100% accurate and hence diagnostic. These unique features helped us formulate an algorithm for preoperative diagnosis of this uncommon form of anorectal malformation in girls seen in North India.

Conclusions: The characteristic features on clinical examination should alert one to the presence of CPC in the outpatient clinic. The AXR was diagnostic in 100% of cases and is mandatory. If any doubt persists, examination of the genitalia under anesthesia with more retraction of the labial folds and endoscopy can be performed for confirmation. These measures should enable a clinician to make an accurate preoperative diagnosis in every girl with CPC.

Keywords: *Anorectal malformation, congenital pouch colon, double vagina, urinary incontinence*

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Introduction

Congenital pouch colon (CPC) is an unusual abnormality in which a pouch-like dilatation of a varying length of the shortened colon is associated with an anorectal malformation (ARM).[1,2] This condition has most frequently been reported from centers in northern India. CPC has been classified into four subtypes (Types I–IV) based on the length of normal colon proximal to the colonic pouch.[1]

The anomalous anatomies in girls with CPC have described the colonic pouch fistula opening either into the vagina or in a persistent cloaca,[1-10] as a colovestibular fistula (colo-VF),[7-13] a colovesical fistula,[5-9,11,14] or even a colouterine fistula.[9] A clearly discernible pattern in the abnormal anatomy was never described. However, two detailed studies reported from our center suggested that these girls did not have a "persistent cloaca" in the accepted sense of the term, as the external genitalia were essentially normal with a wide posterior margin of the vestibule and association

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with spinal or other major malformations was rare.[15,16] All patients had double vagina with monocornuate uterus on each side of the pelvis and a high incidence of urinary incontinence (UI) (75%–81%) due to a widely open bladder neck, a small urinary bladder, and a short, wide urethra.[15,16] The terminal fistula of the colonic pouch was closely apposed to the trigone and urethra and opened into the urethra or the vestibule in either a "high" or a "low" (more distal) intervaginal position.[15,16] Similar findings have been described in a few other reports.[4,5,11,12,17-21] Although there is a spectrum, the findings in our previous studies appeared to be constant and invariable.[15,16] In addition, a preliminary abdominal X-ray (AXR) including the pelvis strongly indicates toward the presence of CPC.

The presence of a single perineal opening on preliminary examination on light labial retraction in those with a "high" confluence of the urethral and vaginal openings can however still lead to a misdiagnosis of persistent cloaca.[16] Girls with CPC and a colo-VF can also mistakenly be diagnosed as having ARM with rectovestibular fistula (recto-VF).[22] Another important feature is that a large number of girls with CPC may present beyond infancy if evacuation is fairly effective through the fistula.[2,6-8,10]

As the management protocol of CPC is vastly different, it is essential to diagnose the anomaly preoperatively. The aim of the present study is to define the important predictive findings and formulate an algorithm to diagnose this anomaly with high accuracy preoperatively.

Materials and Methods

In this retrospective study, the records of 47 girls with CPC managed at our center over a 22-year period (1996–2018) were reviewed. The patients were divided into two groups according to the age at presentation or availability of records: Group A (newborn to 12 months; n = 26) and Group B (>12 months to 20 years; n = 21). The subtypes of CPC were Type I (n = 11; 23.4%), Type II (n = 27; 57.44%), and Type III CPC (n = 6; 12.76%). In three patients, the subtype of CPC had not been recorded as they left the hospital before surgery, but the clinical and radiologic features were diagnostic of CPC. Examination in the outpatient department (OPD) supplemented by an examination under anesthesia (EUA) allowed for comprehensive evaluation. We analyzed the occurrence of various features which appeared vital for diagnosis and formulated an algorithm for accurate diagnosis preoperatively.

Results

In Group A patients (n = 26) [Table 1], important clinical findings were abdominal distension, often marked (n = 14; 53.8%), frequent passage of small amounts of stools and gas from the perineal opening, partial or complete UI (n = 6; 23%), a suspicion of UI (n = 2; 7.7%), and severe perineal excoriations (n = 5; 19.2%). Three "characteristic" features were noted on preliminary examination of the external genitalia and perineum in OPD: (1) a wide posterior margin of the vestibule in all (100%) [Figure 1], in combination with either of these two, (2) separate openings of the urethra and double (often "tilted") vagina with a wide intervaginal bridge; usually showing vaginal mucosa clearly on gentle retraction of the labial folds [Figure 2a]; this was seen in OPD in only 9 (34.6%) patients, or (3) the urethra and double vagina were confluent cranially in 17 patients (65.4%) through a "single opening" seen on gentle retraction in OPD examination, with folds radiating inward from the margins of the vulval vestibule to the point of confluence[15] also described as the "clover leaf" appearance [Figure 2b];[6] this confluence could be appreciated very well with greater retraction of the labial folds possible on EUA, without any need for genitoscopy. The urethral and vaginal confluence was at a more cranial level in 12 patients (46.2%) [Figure 2], while an intermediate picture was present in the remaining 5 girls (19.2%). The colonic pouch ended in a "low" colo-VF (n = 5) [Figure 3], an anterior perineal fistula just posterior to the double vaginas (n = 1), while in 20 patients, the opening of the fistula could not be identified. A fourth peculiar feature noted in 4/7 (57.1%) where photographs were available in records was stretched-out perineum receding inward just posterior to the vestibule [Figure 2a]. AXR was available in 21 patients (81%); it showed either of the two findings in all cases (100%) – a large gas shadow or an air–fluid level (AFL) in the left side of the abdomen (n = 16; 76.2%) [Figure 4a] or a large gas

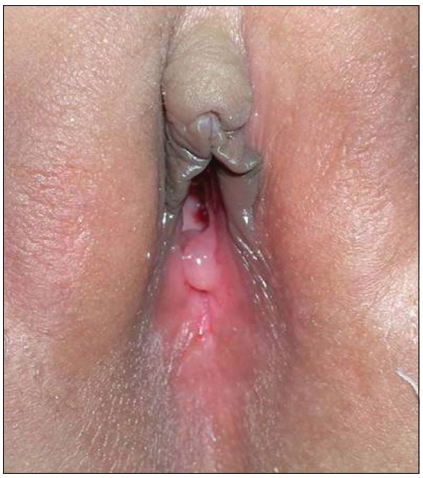

Figure 1: Normal appearance of external genitalia with "wide posterior margin of vestibule"

Table 1: Summary of our findings in Group A (newborn to 12 months) and Group B: >12 months to 20 years

Predictive features	Group A (*n*=26), *n* (%)	Group B (*n*=21), *n* (%)
Clinical findings		
Marked abdominal distension	14 (53.8)	2 (9.52)
UI	6 (23)	18 (85.7)
Suspicion of UI	2 (7.7)	Nil
Severe perineal excoriations	5 (19.2)	5 (23.8)
Examination of external genitalia (including EUA if performed)	26 (100)	21 (100)
"Characteristic features"[15]		
Confluence of urethral opening and openings of double vagina[15,16]		
"Low"	9 (34.6)	13 (62)
"Intermediate" / "Cloverleaf" appearance[6]	5 (19.2)	5 (23.8)
"High"	12 (46.2)	3 (14.3)
Terminal fistula of colonic pouch		
"Low" colo-VF just posterior to urethral opening	5 (19.2)	6 (28.6)
Colo-VF more cranial or "high" level but just posterior to urethral opening	2 (7.7)	2 (9.5)
Anterior perineal fistula just posterior to double vaginas	1 (3.8)	Nil
Distal urethral fistula just within urethral meatus	Nil	2 (9.5)
Fistula into proximal urethra (on endoscopy)	Nil	3 (14.3)
Not identifiable	18 (69.2)	8 (38.1)
Plain X-ray abdomen	21 (81)	18 (85.7)
Large gas shadow with AFL left side of the abdomen	16 (76.2)	8 (44.4)
Large gas shadow with ispissated fecal matter left side of the abdomen	5 (23.8)	10 (55.5)
Pneumoperitonem with large AFL	1 (4.8)	Nil
Normal sacrum	20 (95.2)	18 (100)
Partial sacral agenesis	1 (4.8)	Nil

n: Number of patients with the finding, UI: Urinary incontinence, EUA: Examination under anesthesia, VF: Vestibular fistula, AFL: Air-fluid level

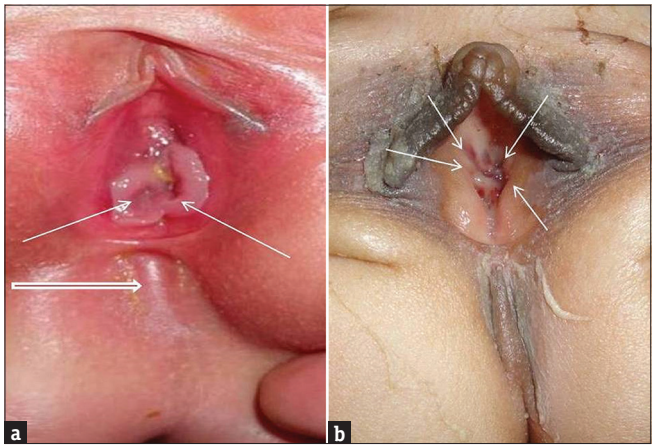

Figure 2: (a) "Low" confluence of openings of the urethra and the two vaginas (fine arrows) in a 3-month old girl with Type II congenital pouch colon. Note the recession in area of perineum posterior to vestibule (broad arrow). (b) External genitalia in a 2-day-old girl with Type I congenital pouch colon showing "clover-leaf" appearance of folds radiating inward and cranially from the margins of the wide vestibule (arrows) suggesting high confluence of urethral and vaginal openings

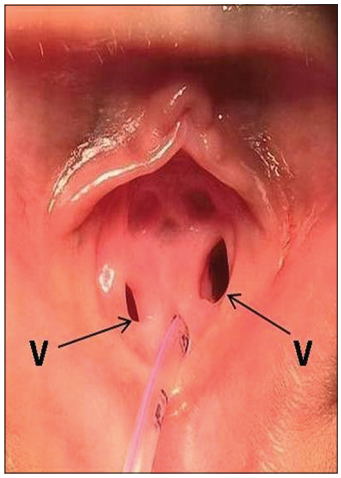

Figure 3: External genitalia in a 6-month-old girl with Type III congenital pouch colon and a "low" colovestibular fistula (catheter). Note the two vaginas (V) with wide intervaginal bridge

shadow with inspissated fecal matter on the left side abdomen (*n* = 5; 23.8%). In one patient, in addition to the large AFL, pneumoperitoneum due to perforation of the colonic pouch was also present. Sacrum was normal in 20/21 (95.2%) patients [Figure 4a] and partial sacral agenesis was seen in one patient.

In Group B patients (*n* = 21) [Table 1], at presentation, abdominal distension was present in two patients (9.5%); and extensive peri-vulval excoriations were noted in five patients (23.8%) [Figure 5a]. In six patients (28.6%), the abdomen had a doughy feel on palpation, perhaps due to the fecaloma in the distended colonic pouch. UI was present in 18 patients (85.7%); of these, partial UI

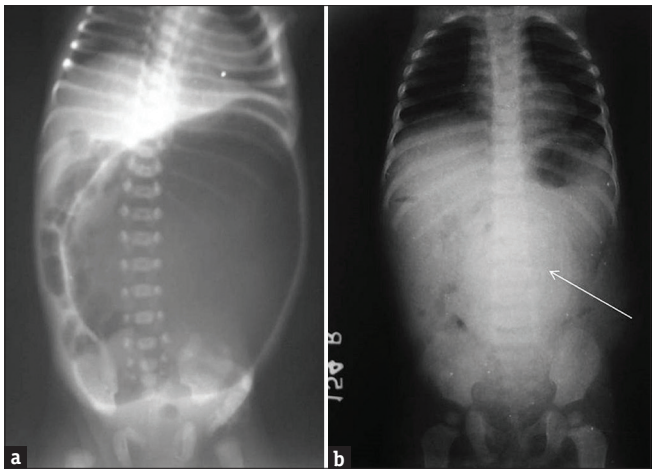

Figure 4: (a) Abdominal X-ray in a newborn girl with Type II congenital pouch colon showing a large left-sided abdominal gas shadow. Note the normal sacrum. (b) Abdominal X-ray in a 2-year-old girl with Type I congenital pouch colon showing a large radio-opacity on the left side of a distended abdomen due to inspissated, accumulated fecal matter (arrow)

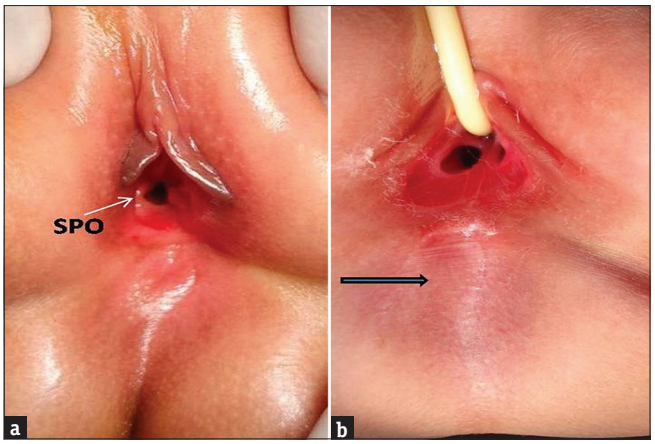

Figure 5: (a) A 15-month–old girl with Type II congenital pouch colon showing a single perineal opening. (b) On retraction of labial fold, the openings of the urethra and vaginas. Arrow points to inwardly receded perineum

in 5 (23.8%) and complete UI in 13 patients (61.9%) were seen. Examination showed characteristic features of the external genitalia described above in Group A in all 21 patients (100%). OPD examination showed only a single, though wide, perineal opening in 8 of these 21 patients (38.1%) [Figure 5a]; the confluent urethral and vaginal orifices could be appreciated well with greater retraction of the labia on EUA without any need for genitoscopy [Figure 5b]. In older patients, a wide intervaginal bridge was more apparent between the two vaginal openings [Figure 6a and b]. The fistulous opening of the colonic pouch could be seen in ten patients (47.6%); it was closely apposed to the trigone and urethra and opened posterior to the urethral opening in seven patients – "low" (intervaginal) vestibular fistula (VF) in five [Figure 6a] and "high" or more cranially placed VF in two. The fistula opened into the distal urethra just within the wide urethral meatus in three patients [Figure 6b] and into the proximal urethra in another three patients on cystourethrogenitoscopy. The opening of the fistula was not identified in the records of 8 patients (38.1%). In addition, stretched-out perineum receding inward just posterior to the vestibule was noted in 6/11 patients (55.55%) in the available photographic records [Figure 5b]. AXR was available in 18 patients; all showed either a gas shadow or AFL mostly in young patients or a large radio-opacity on the left side of a distended abdomen due to fecaloma, especially in older girls and a normal sacrum [Figure 4b]. With these features, a preoperative diagnosis of CPC had been made in 17/24 (70.83%) girls with CPC in the last 10-year period from 2009 to 2018. An algorithm to maximize chances of preoperative diagnosis is shown in Table 2.

Discussion

Although clinical features like marked abdominal distension in newborns and infants and poorly managed constipation with overflow incontinence in older girls are important findings suggesting the possibility of CPC in girls with ARM, the characteristic features on examination of the external genitalia are quite unique.[15,16] A history of UI is an additional feature pointing to the diagnosis.[16] Often, in the newborn or infant in diapers, the presence of UI may be missed initially, especially as UI may be partial and the child would also be passing urine intermittently in a stream. However, as in 2 infants (7.7%) from the present study, careful questioning may elicit that the parents have noticed leakage of urine when the child strains or sometimes even in the standing posture. UI is more clearly apparent in older girls[15,16] and in the present study, a history suggestive of UI was present in 18/21 (85.7%) older girls and only in 8/26 (30.6%) newborns or infants. Apart from the characteristic features of the external genitalia, peri-vulval excoriations due to UI may also be present.[15,16] In older girls, the underlying inspissated fecal matter in the large colonic pouch gave a doughy feel to the abdomen on palpation.

As shown by the algorithm for diagnosis [Table 2], AXR findings are usually conclusive, even in older girls in whom a large radio-opacity on the left side of a distended abdomen due to inspissated accumulated fecal matter in the colonic pouch is often a very characteristic finding. As described earlier, at least in reports from the Indian subcontinent, despite behaving as a high type of ARM, CPC is infrequently associated with sacral anomalies or major malformations involving other organ systems.[23,24] In the present study, a normal sacrum was present in 38/39 patients (97.4%) and

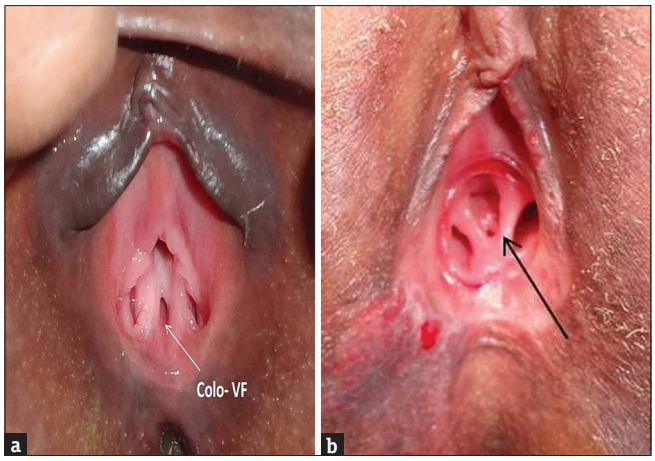

Figure 6: (a) External genitalia in a 10-year-old girl with Type II congenital pouch colon and distal colo-urethral fistula (arrow). Note wide urethral meatus, wide inter-vaginal bridge, and extensive "peri-vulval excoriations."Reproduced with permission from Figure 7 in Chadha and Khan.[24] (b) External genitalia in a 12-year-old girl with Type II congenital pouch colon and a "low" colo-vestibular fistula. Note the wide intervaginal bridge

Table 2: Algorithm for preoperative diagnosis of congenital pouch colon in girls

History and Examination in OPD *["Characteristic" appearance of external genitalia and perineum]* *Spine normal*
STRONG SUSPICION OF CPC
Plain X-Ray Abdomen *Large gas shadow +/- AFL or inspissated stools left side abdomen* *Normal Sacrum*
VIRTUALLY CONFIRMATORY : 100 % IN OUR SERIES
Examination of external genitalia under anesthesia (EUA) +/- Cystourethrogenitoscopy Contrast study through VF if present and accessible *(rarely required)*
CONFIRMATORY

CPC: Congenital pouch colon, OPD: Outpatient department, EUA: Examination under anesthesia, VF: Vestibular fistula, AFL: Air–fluid level

this finding is also an important pointer toward the diagnosis.

A colo-VF is often present in girls with CPC and may be visible on initial examination or at EUA as in 19/47 (40.4%) patients in this series. In cases of CPC with a low confluence of the perineal openings, an erroneous diagnosis of ARM with recto-VF is therefore very much possible, especially as recto-VF is also associated with a septate vagina in as many as 6% cases.[22] In addition to the important differentiating features described earlier, in 55.55% (Group A = 4/7, Group B = 6/11) girls in our series, the whole perineum appeared receded inward, and the area of the anal dimple appeared wide and stretched out. Similar findings reported earlier[25,26] is an additional diagnostic feature.

The differentiation of CPC from persistent cloaca is also important as the management protocol for both conditions is vastly different. The external genitalia are hypoplastic in cloaca, contrary to grossly normal appearance with wide posterior margin of vestibule in CPC. Cloacal anomalies are often associated with major GU anomalies including hydrocolpos, sacral anomalies, and major malformations involving other major organ systems;[22,27] these are characteristically absent in CPC.[24] In addition to the preliminary colostomy for the ARM, associated urologic anomalies in cloaca require detailed assessment and may need emergent management.[22]

The importance of distinguishing CPC with a colo-VF from recto-VF cannot be overemphasized. Recto-VF is usually managed by a single-stage procedure, limited PSARP, in infancy[27,28] although a preliminary colostomy is recommended for certain specific indications.[27] If the presence of CPC is undiagnosed preoperatively, the procedure performed would be fraught with complication as the colo-VF tract is closely apposed to the urethra and trigone in an intervaginal position[15,16] and the consequences of damaging these structures and/or the two vaginas may be serious. In addition, management of the colonic pouch is in itself an additional necessity. Agarwal *et al*. reported a case of CPC with a VF misdiagnosed as a recto-VF in whom initial limited PSARP for the VF was followed by wound disruption. Type I CPC was detected only during a diverting colostomy and the child ultimately required a three-stage reconstruction for the anomaly.[28]

It has been suggested that in girls with a VF who do not decompress well despite a wide fistula orifice, a barium enema should routinely be performed to rule out CPC.[3,29] In our view, a contrast enema through a VF, if present, should rarely be required as the diagnostic protocol described here [Table 2] should enable preoperative diagnosis of CPC. Perhaps, the only indication should be the rare older girl with a colo-VF in whom the typical features of the external genitalia, perineum, anal dimple, and AXR findings are inconclusive.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient (s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initial s will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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