

Diagnostic suggestion and surgical consideration for Hirschsprung's disease associated with high anorectal malformation

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Object The objective of this study were to highlight the finding of vasa recta (tortuous) on the colonic wall as a diagnostic clue for suspecting an associated Hirschsprung's disease (HD) and to draw attention to the importance of preserving the aganglionic rectum plus a retrorectal pull through in these cases.

Background The association of HD with anorectal malformation (ARM) is both diagnostically and surgically challenging.

Patients and methods Records of cases with ARM treated over 15 years were examined. Among these, five children with an associated HD were analyzed with regard to their clinical, radiological, surgical, histopathological findings, and outcome. Relevant literature was also reviewed.

Results HD was present in 1.26% cases of ARM. All were men with high-type ARM. Two groups were identified. In group 1 (two patients), associated HD was suspected after completion of all stages of ARM repair. They reported prolonged postoperative constipation, abdominal distention, and enterocolitis. Moreover, they endured additional surgeries for HD. In group 2 (three patients), HD was suspected at the time of initial colostomy for ARM in

two patients on visualizing tortuous (cork screw) vasa recta on the sigmoid colon surface in a region similar to the transition zone in HD. Biopsy from the site confirmed HD. In the third patient, these vessels were visualized on the colostomy loop at the time of laparoscopic pull through for ARM. Preservation of the aganglionic rectum (to function as a fecal reservoir) as well as a retrorectal pull through was done in both groups.

Conclusion Finding prominent corkscrew vessels on the colonic surface could serve as a clinical clue for the presence of HD in cases of ARM. Moreover, preserving the aganglionic rectum and performing a Duhamel pull through helps provide acceptable continence. *Ann Pediatr Surg* 14:78–82 © 2018 Annals of Pediatric Surgery.

Annals of Pediatric Surgery 2018, 14:78–82

Keywords: anorectal malformation, Hirschsprung's disease, rectal reservoir, tortuous vasa recta

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Received 1 February 2017 accepted 15 April 2017

Introduction

The reported incidence of Hirschsprung's disease (HD) in patients with anorectal malformations (ARM) is between 2.3 and 5.6% [1–3]. This association poses both a diagnostic as well as a surgical challenge. Symptoms attributed to ARM repair mask the underlying HD in both low and high ARM and the stoma for high ARM is usually fashioned proximal to the aganglionic segment [4]. We aim to highlight our finding of an aberrant 'corkscrew' vasa recta observed on the colonic surface in the area of the transition zone. The finding could act as a clinical clue for suspecting an associated HD in cases of high ARM. Furthermore, we suggest that one can provide acceptable continence in these cases if one preserves and places the aganglionic rectum within the available sphincter complex (as a fecal reservoir) and then performs a retrorectal (Duhamel) pull-through of the proximal ganglionic segment.

Patients and methods

The records of all 398 patients with ARM admitted from August 2001 to December 2015 were checked. Of these, five male patients with ARM had associated HD. Their record was evaluated with respect to clinical history, physical examination, radiological findings, surgical

procedures performed, surgical/histopathological findings, and outcome. In these five cases, HD was suspected on the basis of their clinical presentation/course, intraoperative, or radiological findings (Table 1).

In two cases (patients 1 and 4), HD was suspected following the completion of all three stages of ARM repair. They developed constipation and abdominal distension following colostomy closure that did not respond to conventional management. In these patients, a contrast enema film was also obtained to confirm the diagnosis. Computed tomography of pelvis was performed in one patient to document the placement of the rectum within the sphincters. In addition, full-thickness rectal biopsies (additional stomal biopsy in one case) were taken to document aganglionosis in these cases.

In the remaining three cases, HD was suspected intraoperatively either at the time of neonatal colostomy for ARM (patients 2 and 3) or at the time of pull-through for ARM (patient 5). In the former two, a gradual narrowing in the sigmoid colon akin to a transition zone in HD led to the suspicion. Here, we also observed the presence of abnormal colonic vasa recta in the area of the transition zone (described later). In patient 5, HD was suspected on visualizing these abnormal vasa recta on the

Table 1 Clinical and management summary of the five cases

Case numbers	Initial diverting stoma	Pull-through/age	Age at colostomy closure	Age at first suspicion of HD	Features suggesting the possibility of associated Hirschsprung's disease	Contrast enema findings	Biopsy site/ biopsy report	Surgical procedure/age	Total procedures	Follow-up duration/clinical condition
1	Transverse colostomy (Done outside)	PSARP 4 months	8 months	23 months (15 months after transverse colostomy closure)	Recurrent abdominal distension, constipation, visible bowel loops, soiling, loud borborygmi. Dilated colon on X ray film	Transition zone at Rectosigmoid	Rectum – ganglion cell absent	Single stage Duhamel pull-through. 9 cm of very dilated proximal normal colon resected along with aganglionic bowel	4	Up to 3 years No fecal soiling, incontinence/enterocolitis
2	Midsigmoid	PSARP 9 months	Not done	At initial colostomy (day 1 of life)	Presence of prominent corkscrew-shaped vessel at the region of mid sigmoid associated with a change in bowel caliber	Not done	Stoma site – absent ganglion cells. 5 cm. Proximal to stoma site – ganglion cells present	Martin's modification of Duhamel at 3 years. Pull-through of the proximal ganglionic colon	3	Still in follow-up Hypospadias repair done No fecal soiling or enterocolitis
3	High sigmoid	Laparoscopic-assisted pull-through 4 months	Not done	At initial colostomy (day 2 of life)	A change in bowel caliber observed 2 cm above the rectosigmoid junction. Corkscrew vessel observed in this area	Not done	Rectosigmoid junction – absent ganglion cells. Colostomy site – positive for ganglion cells	Laparoscopic-assisted Duhamel at 7 months	3	2 years One episode of enterocolitis No fecal soiling
4	Completed all three stages of ARM elsewhere. A repeat sigmoid colostomy was performed before referral			24 months	Persistent constipation and recurrent distension. Three episodes of enterocolitis following ARM repair. Rectum seen within the sphincters on CECT pelvis	Done before referral showed the transition zone at the rectosigmoid	Rectum – absent ganglion cells Stoma site – ganglion cells present	Duhamel pull-through 5 cm of dilated colon proximal to the redo stoma site excised. Required mobilization of splenic flexure	5	Still in follow-up Fecal soiling 2–3 per week No enterocolitis
5	High sigmoid	Laparoscopic-assisted pull-through 3 months	Not done	3 months At the time of pull-through for ARM	Corkscrew vessels observed at the time of laparoscopic pull-through for ARM in both proximal and distal loops adjacent to the stoma site.	Not done	Stoma site – absent ganglion cells 5 cm. proximal to the stoma site – ganglion cells present	Laparoscopic-assisted Duhamel at 6 months	3	2 years No fecal soiling One episode of enterocolitis

ARM, anorectal malformations; CECT, contrast-enhanced computed tomography; HD, Hirschsprung's disease; PSARP, posterior sagittal anorectoplasty.

colonic wall at the time of laparoscopic pull-through for ARM. Even though there was no evidence of a transition zone in this case, on the basis of our experience in patients 2 and 3, we suspected HD. Intraoperative full-thickness colonic biopsies were obtained in these three cases from relevant sites accordingly. The histological examination was performed under hematoxylin and eosin staining. We do not routinely perform biopsy for HD in every case of ARM. The patient's follow-up was recorded up to his last outpatient clinic visit.

Result

Out of the 398 cases of ARM, HD was diagnosed in five (1.26%) cases. All were males with high ARM. Three had a rectobulbourethral fistula, one had an anorectal stenosis, and in one (patient 4) the type of high ARM was not known. Broadly, there were two distinct groups of patients.

Group 1 ($n = 2$): in these children, the diagnosis of an associated HD was suspected after the completion of all stages of ARM repair (patients 1 and 4). Their age at suspicion of HD was 23 and 24 months. These two patients had constipation, abdominal distension, decreased somatic growth, chronic use of enemas, and recurrent enterocolitis.

Group 2 ($n = 3$): in these children, HD was diagnosed either at the time of initial colostomy (patients 2 and 3) or at the time of pull-through for ARM (patient 5). They did not have any major problems apart from those related to colostomy. An associated tracheoesophageal fistula and a penoscrotal hypospadias were present in one patient each. A brief summary of the five cases and their management is presented in Table 1.

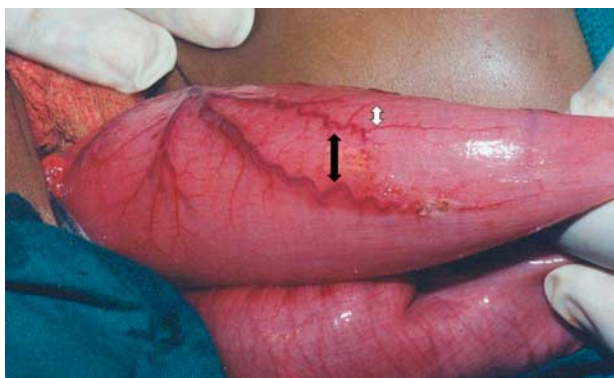
In the patients in group 2, we identified the presence of an altered pattern of vessels (colonic vasa recta) in the

subserosal plane of the sigmoid colon. In two babies (patients 2 and 3), this pattern was observed at the time of initial colostomy in the region of the visible transitional zone (Fig. 1), whereas in patient 5, these vessels were visualized at the time of laparoscopic pull-through for ARM (Fig. 2). The vasa recta appeared prominent, thickened, lengthened, tortuous (corkscrew shape), and comparatively darker in color than those in the adjoining normal loop. The vessels followed an oblique course nearly parallel to the long axis of the bowel in contrast to the normal vasa recta, which course perpendicular to the long axis (Fig. 1). In the magnified laparoscopic view (Fig. 2), the colonic vasa brevia were also observed to be tortuous. It was observed that despite an initial diverting colostomy performed for ARM in patients 2 and 3, the tortuosity persisted till the time of pull-through.

Discussion

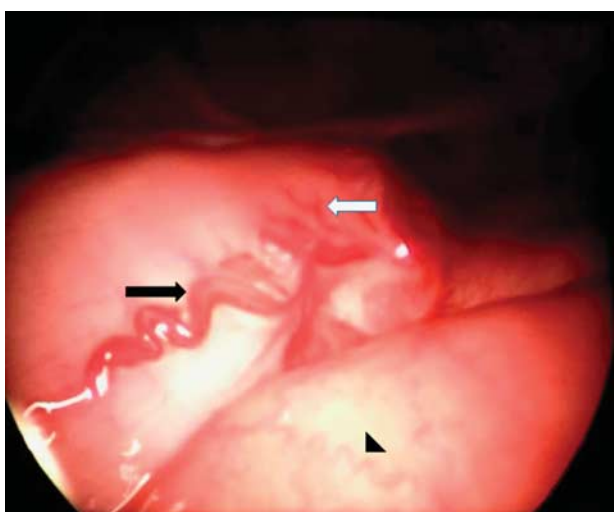
The reported incidence of associated HD in cases of ARM lies between 2.3 and 5.6% [1–3]. In a recent systemic review of 38 articles (90 cases), the reported

Fig. 1



Tortuous vasa recta (black double arrow) and its branches (white double arrow) in the region of the transition zone.

Fig. 2



Tortuous vasa recta (black arrow), vasa brevia (white arrow) observed in the distal stoma loop. Tortuosity was also observed in the proximal stoma loop (black arrow head).

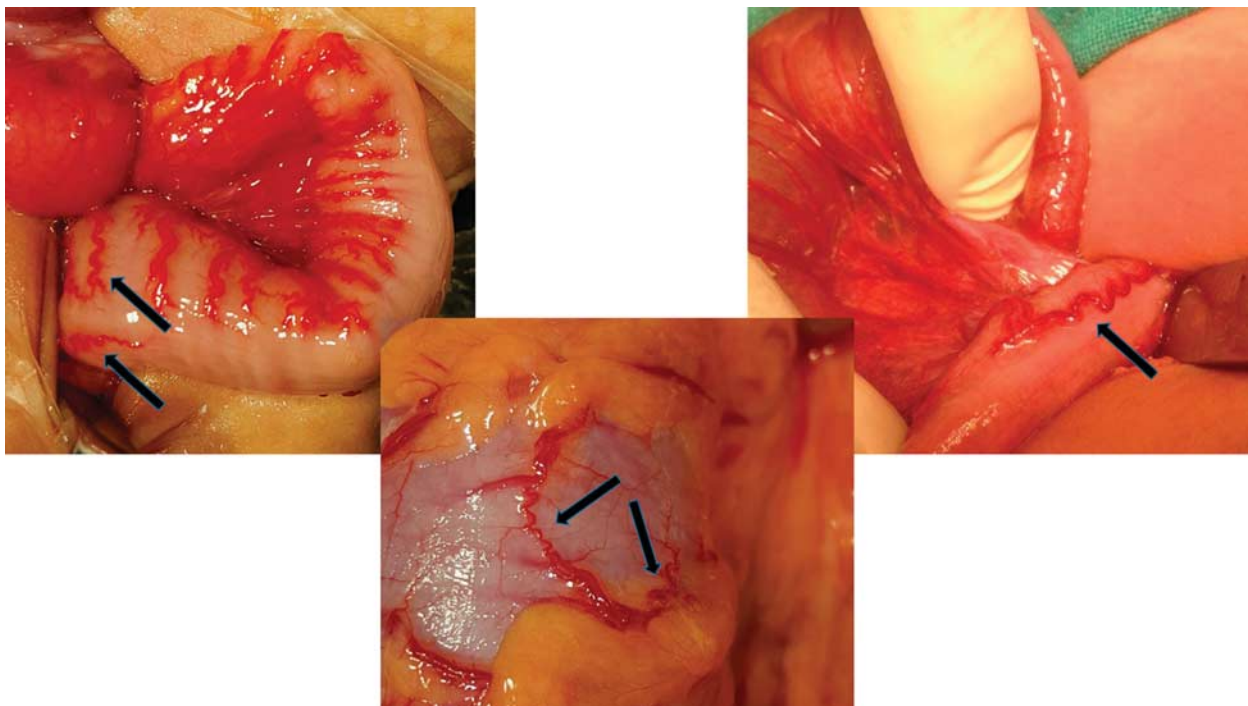
incidence was 2% [4]. In our series, it is 1.26% (5/398 cases). Nearly 35% of the initial colostomies performed in patients with ARM with HD are created in the aganglionic segment and in nearly 60% of the cases, the diagnosis of HD is not known at the time of operative ARM correction, leading to a median delay of 8 months for the diagnosis of HD from the initial diagnosis of ARM and also a median delay of 17.5 months in the operative correction of HD in these patients [4]. Conversely, anything that points toward a possible association of HD in cases of ARM can help reduce the child's morbidity as well as the surgeon's difficulty.

The finding of an aberrant vasculature in patients with HD was first reported by Lister [5] in 3/10 patients in his series. He described these vessels as firm, thick, and running in the mesentery of the colon, most conspicuously in the junctional zone. Another series reported abnormal arteries in 20 of 62 cases with HD at microscopic examination [6]. They were mostly located in the histological transition zone and were cited in the submucosa, muscle layer, and subserosa in 100, 6, and 18% of cases, respectively. The authors hypothesized that this abnormal pattern could expand proximally up to the mesenteric vessels as observed by Lister and that this histological finding was consistent with 'adventitial fibromuscular dysplasia' described by Stanley *et al.* [7]. Histologic abnormalities in the smooth muscle actin filament along with increased collagen fiber expression around the submucosal vessels of large bowel biopsies in more than 60% of cases of intestinal neuronal dysplasia (IND), IND with HD, have been reported [8] and advocated as an additional diagnostic feature in cases of IND. Grossly abnormal tortuous vessels have also been reported in the segmental dilatation of the intestine [9–13].

Similar to Lister, we also observed tortuosity of the vessels in the area of the transition zone in cases of isolated HD, but unlike Lister, who reported these vessels in the adjoining mesentery of the transition zone, we observed these on the bowel wall (Fig. 3). A similar pattern of vessels cited in cases of ARM in a region similar to the transition zone in HD led us to take a biopsy from that area. The biopsy specimens from these sites were aganglionic. Furthermore, we observed that the tortuosity in these vessels persisted till the time of pull-through in patients 2, 3, and 5 even after the creation of a defunctioning stoma in these cases. This implies that the tortuosity was not secondary to the congenital bowel obstruction, but rather an inherent developmental abnormality of these vessels. We did not observe such vessels in our group 1 patients. We believe that owing to the previous pull-through for ARM performed in these patients, the concerned involved region part moved further down in the pelvis and became clinically difficult to visualize. In addition, the postoperative fibrosis can also alter the picture.

In the patients in group 1, the diagnosis of an associated HD was suspected following complete repair of ARM. This increased the total number of surgeries in these children to 4 and 5, respectively (Table 1). In the

Fig. 3



Rectosigmoid in cases of Hirschsprung's disease showing tortuous vasa recta (black arrow) in the region of the transition zone.

patients in group 2, the diagnosis of aganglionosis was known before pull-through for ARM. Here, the total number of surgeries remained three. For the patients in group 2, one can enquire as to why a simultaneous correction of both ARM and HD was not performed. Considering that 60% of the time the diagnosis of an associated HD is not known at the time of ARM repair, only 10 cases are reported on simultaneous pull-through for both ARM and HD [2,14–17]. Surprisingly, the postoperative outcome is reported only in 4/10 patients, of whom two were females with a low ARM [17].

We believe that cases of high ARM associated with HD are more complex to treat. These patients have an absent anus, an aganglionic rectosigmoid of varying length, and a malformed sphincter mechanism. Excision of the aganglionic segment and bringing down the normal ganglionic (proximal sigmoid or descending colon) as a neoanus in these patients would be akin to a perineal colostomy. Thus, to improve continence, preservation of the distal bowel (rectum) as a reservoir is essential. A subsequent retrorectal (Duhamel) procedure involves minimal burden for an already compromised anal sphincter and bladder innervations [18]. On similar grounds, both our patients in group 1 were also treated with a retrorectal pull-through, preserving the previously pulled-through aganglionic segment. A timely diagnosis of an associated HD and two sequentially staged pull-through procedures limited the surgical procedures to three in our (group 2) patients. Moreover, a retrorectal pull-through helped in achieving acceptable continence levels in all our patients. Our data are limited for us to make a generalized statement, but then the worldwide incidence of this association is itself very low to make a meaningful

Table 2 Clinical, radiological, and surgical findings reported in the literature suggesting Hirschsprung's disease in patients with anorectal malformations

Clinical/radiological finding	Reported cases	Total cases
At the time of neonatal colostomy		
Narrowing/change in the caliber of the colon [2]	1	
Following neonatal colostomy		
Nonfunctioning proximal stoma [2,3]	4	9
In distal cologram film		
Narrowing/change in the caliber of the colon [1,21]	3	
At colostomy closure		
Grossly dilated proximal bowel in a properly placed nonstenosed stoma [22]	1	
Following the completion of all stages of ARM repair		
Abdominal distension, vomiting, recurrent constipation, enterocolitis despite confirmation of a properly placed normal-caliber anus. [1,3,18,21]	11	18
Transition zone observed on barium enema [18,21]	2	
Poor evacuation of dye after barium enema [1]	5	

ARM, anorectal malformations.

comparison from the available literature. Moreover, to date, the use of Duhamel pull-through in these cases has been reported only once [18].

Up to 25% of cases of ARM with HD reportedly have a syndromic association, namely, (Currarino syndrome, Down's syndrome, cat-eye syndrome) [2,4] and Pallister–Hall syndrome [19]. In addition, it is reported in 17% cases of ARM with coloboma of the iris [20]. We did not have any apparent syndromic association in our cases. Other reported clinical, radiological, and surgical pointers in the literature suggesting HD in patients with ARM are presented in Table 2. With these clinical clues,

an associated HD was detected before the completion of ARM repair only in nine cases, whereas 18 cases were detected after ARM repair. Nevertheless, we suggest that siting of corkscrew colonic vasa recta in cases of high ARM, particularly in a segment resembling a transition zone, could prompt the surgeon to at least suspect an associated HD and take a biopsy from the site. Timely identification of an associated HD could help the surgeon re-plan his surgery.

Conclusion

The association of HD with ARM can aggravate a patient's morbidity because of the masking of HD and a consequent delayed diagnosis. The finding of (corkscrew shaped) vasa recta on the surface of the colon, particularly if accompanied by a change in bowel caliber, may be a clinical clue for the presence of HD in cases of ARM and prompt a biopsy from the site. Moreover, to improve continence, preservation of the distal aganglionic bowel (rectum), as a reservoir, with a subsequent retrorectal pull-through is beneficial.

Conflicts of interest

There are no conflicts of interest.

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