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How to manage a late diagnosed Hirschsprung's disease

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ABSTRACT

Background: How to manage a late diagnosed Hirschsprung's disease (HD) and how to avoid calibre discrepancy? **Subjects and Methods:** A retrospective study of all patients diagnosed with HD over 2 years in our hospital from January 2009 to December 2012. Data were analysed for clinical presentations, investigations, surgical procedures and post-operative outcome. **Results:** Fifteen patients, operated by one single surgeon, were included in this study. The mean age was 6 years (2-16 years). Patients had an ultra-short segment type in 4 cases, rectosigmoid type in 9 cases and descending colonic aganglionosis in 2 cases. Rectal wash out was effective in 12 patients. A blowhole transverse colostomy was performed in 2 patients. Twelve patients underwent one single stage endorectal pull-through. Anastomosis incongruence was avoided by a plication procedure never described before. The assessment of post-operative outcomes by the paediatric incontinence and constipation scoring system revealed a normal continence function in all our patients, but 3 patients suffered from soiling secondary to constipation. **Conclusion:** One single stage pull-through can be safe and effective in children with late diagnosed HD. Routine rectal washout is a good way to prepare the colon. In some cases, blowhole colostomy can be an option. Anastomosis incongruence is a challenge; we describe a plication procedure to avoid it.

Key words: Anastomosis incongruence, blowhole colostomy, Hirschsprung's disease, late diagnosis, one stage endorectal pull-through, plication procedure

INTRODUCTION

Hirschsprung's disease (HD) is a developmental disorder of the enteric nervous system that is characterized by the absence of ganglion cells in the myenteric and submucosal plexus from the anorectum for a variable colonic or intestinal distance. This results in a lack of peristalsis leading to a huge dilatation of the colon proximal to the aganglionic bowel. Late diagnosed HD is a challenge in developing countries because it may have a negative impact on the management of HD and rule out one stage operative procedure. The bowel in the patients, mentioned above, is chronically dilated and especially the colon proximal to the aganglionic bowel tends to be severely dilated. This is a report of a method to narrow the dilated proximal colon to facilitate one stage pull-through.

SUBJECTS AND METHODS

A retrospective chart review study was conducted to evaluate the clinical records data of cases of HD in children over 2 years who were diagnosed from January 2009 to December 2012. Fifteen children with late diagnosed HD operated by one surgeon were recorded. Their charts were evaluated for clinical presentation, pre-operative management, surgical approaches, post-operative complications and long-term follow-up.

Inclusion criteria

Children over 2 years were diagnosed and operated for HD because the colon proximal to the aganglionic bowel in these patients tends to be severely dilated with an important faecal stasis. All these lead to a difficult

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pre-operative preparation and a challenging one stage endorectal pull-through surgery. Diagnosis is made by a combination of medical history, examination and a barium enema to determine the type of HD using the level of the transition zone. The rectal biopsy is used to give definitive diagnosis. The functional outcome was assessed according to the paediatric incontinence and constipation scoring system (PICSS) developed by Fichtner-Feigl *et al*.[1] Telephone and outpatient hospital interviews using the PICSS were performed with the older patients and the mothers of the others, but 1 patient who moved to another city could not be interviewed.

RESULTS

Eleven males and 4 females were recorded. Ranged age at diagnosis was from 2 to 16 years (with a mean of 6 years).

Table 1 documents the clinical features. 15 patients have had chronic constipation since they were born. Delayed passage of the meconium was reported in 9 patients 48 h after their birth. Hirschsprung-associated enterocolitis (HAEC) at presentation was seen in 6 patients, abdominal distension in 10 patients [Figure 1] and palpation of large faecaloma in 7 patients. Further presenting features were: Malnutrition and failure to thrive in 5 patients and severe anaemia in 1 patient who had required transfusion before surgery. All the patients had a special pre-surgical HD diet and probiotics. No associated anomaly was present.

Table 2 documents radiological finding; plain abdominal X-ray showed a faecal stasis in 11 patients, air fluid levels in 8 cases and a cut-off sign in 4 patients [Figure 2]. After an initial bowel preparation during 3-4 weeks a contrast enema is performed [Figure 3]. The extent of colonic

Table 1: Patient profile and clinical description of the patients

Case *n*	Age (year)	Gender	Delayed passage of meconium	Chronic constipation	Failure to thrive	Gross abdominal distension	Palpable faecaloma	Associated enterocolitis
1	2	Male	—	Yes				
2	2	Female	Yes	Yes		Yes		Yes
3	3	Male	yes	Yes			Yes	
4	3	Female	Yes	Yes		Yes		Yes
5	3	Male	Yes	Yes	Yes			Yes
6	4	Male	Yes	Yes	Yes	Yes		
7	4	Male	Yes	Yes		Yes	Yes	
8	5,6	Male	—	Yes		Yes		
9	6	Male	—	Yes	Yes	Yes	Yes	
10	6	Male	—	Yes				
11	8	Male	Yes	Yes	Yes	Yes	Yes	Yes
12	8	Male	—	Yes		Yes	Yes	Yes
13	9	Male	Yes	Yes		yes	Yes	Yes
14	9	Male	—	Yes	Yes		Obstruction	
15	16	Male	Yes	Yes		Yes	Yes	

Table 2: Radiological finding and pre-operative management

Case	Faecal stasis	Air fluid levels	Cut-off sign	Transition zone	Non-operative bowel preparation (days)	Blow hole colostomy	Ileostomy
1	Yes	Yes		Short	30		
2	Yes			Recto-sigmoid	30		
3	Yes			Short	20		
4		Yes	Yes	Recto-sigmoid	60		
5		Yes	Yes	Recto-sigmoid	60		
6	Yes			Recto-sigmoid	75		
7	Yes	Yes		Recto-sigmoid	60		
8	Yes			Short	75		
9	Yes	Yes	Yes	Recto-sigmoid	30 (disimpacted)		
10				Short	210		
11	Yes	Yes		Descending colon	150	Yes	
12		Yes	Yes	Recto-sigmoid	120	Yes	
13	Yes	Yes		Recto-sigmoid	60		
14	No						Yes
15	Yes			Recto-sigmoid	30		

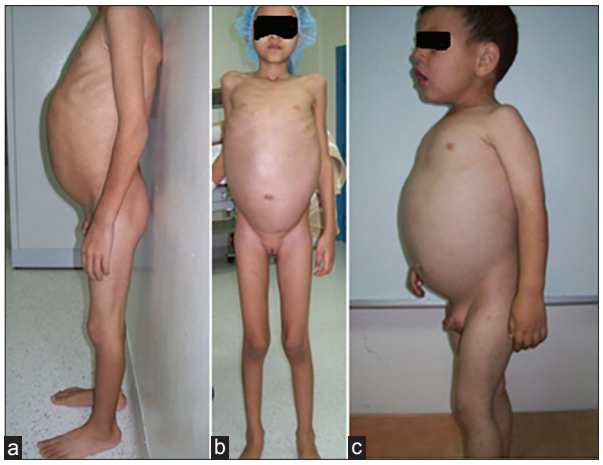

Figure 1: Abdominal distension in late diagnosed Hirschsprung disease in: (a) 8-year-old boy, (b) 8-year-old girl and (c) 3-year-old boy

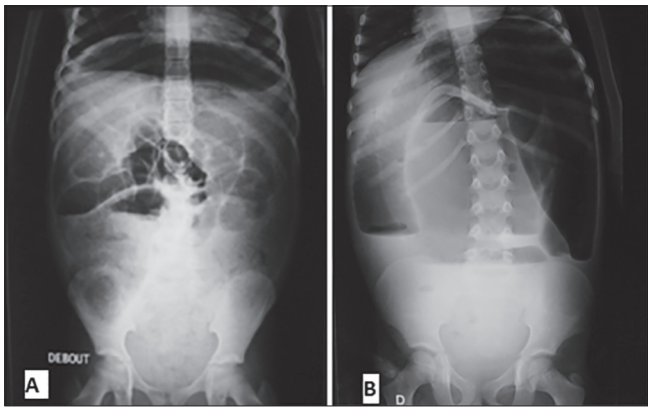

Figure 2: Abdominal X-ray showing: (A) Faecal stasis and huge dilated transverse colon. (B) Air fuid levels and cut-off sign

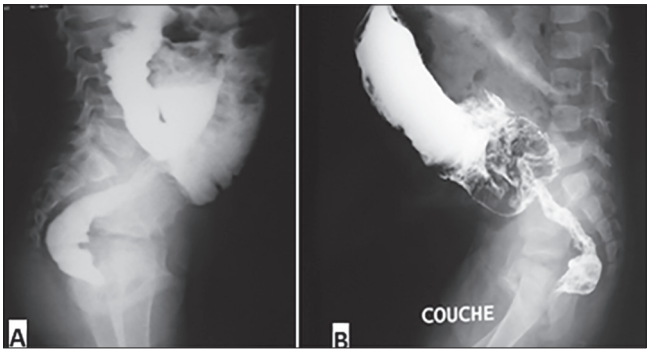

Figure 3: Contrast enema demonstrate in (A and B) a rectosigmoid transition

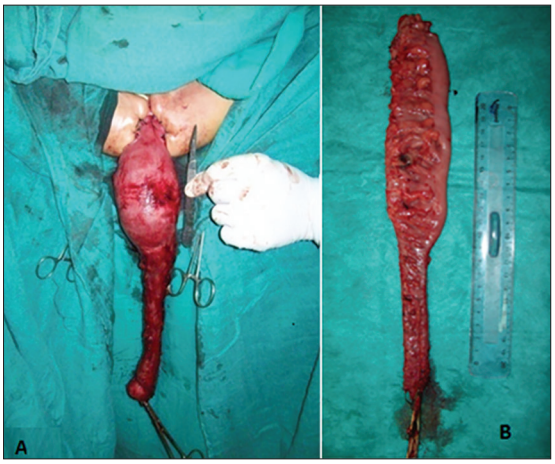

Figure 4: (A) During pull-through we get to a point of calibre change in the colon. (B) After the frozen section documenting ganglion cells, resection of the aganglionic bowel and the severely dilated proximal colon

involvement was defined by the level of transition zone, it showed a short type HD in 4 patients, a rectosigmoid in 9 and a long segment (descending colon) in 2. Anorectal manometry was performed in 6 cases; it showed a lack of normal recto-anal inhibitory reflex. Histology of surgical rectal biopsy under general anaesthesia confirmed the diagnosis of HD in 14 patients, but in the one referred patient this biopsy had been done during a previous laparotomy for obstruction in the referring hospital. The pre-operative bowel preparation by rectal washout was effective in 12 patients, but one of them was disimpacted under anaesthesia. In 2 patients with a huge dilatated colorectal washout was ineffective, so we performed a blowhole transverse colostomy followed by an immediate decompression. An ileostomy was performed during the previous laparotomy for obstruction in the referred patient. The mean time of bowel preparation was 67 days (20-210 days). The surgical procedure was one stage transanal endorectal pull-through (OTEPT) in 12 patients [Figure 4]. Three other patients underwent a multistage endorectal pull-through assisted by laparotomy in 2 patients with the closure of blowhole colostomy and the referred patient with a long aganglionosis had Deloyers procedure with the closure of ileostomy. Dissection was difficult in older children. After extemporaneous analysis of biopsies in the normal colon, a resection of the aganglionic bowel in addition to the remaining dilated colonic segment performed then followed by coloanal anastomosis. Anastomotic incongruence was present in all those patients and was avoided by a plication procedure. This procedure began by four quadrant sutures followed by four plicating sutures performed in between each two quadrant sutures; we finished this anastomosis by one stiche between each couple of the previous quadrant suture and the plicating suture. At the end, a total of 16 stiches are sufficient for anastomosis and for reducing calibre discrepancy [Figure 5]. No intra-operative complications or bleeding were encountered. Normal bowel movement was: 12-24 h in 6 patients, 24-48 h in 4 patients and more than 48 h in 5 patients. The mean hospital stay after surgery was 8 days (3-15 days). Histological examination of the operative specimen confirmed the diagnosis of HD without residual disease in the proximal colon in all patients. A weekly dilatation

was performed 3 weeks after surgery during 8 weeks if needed. Three patients had perineal excoriation in the early post-operative period, and anastomotic stricture in 3 patients treated successfully by dilatation. Table 3 documents the post-operative results, with a median 2, 6 years follow-up: Four patients were reported with HAEC, only one of them had a history of HAEC before surgery. The incontinence scores in the present series were from the 22.5 to 28. The incontinence scores were superior to their age-specific lower limit 95% confidence interval (CI) PICSS scores. Three patients (number 6, 10 and 13) had scored constipation below their age-specific lower limit 95% CI PICSS scores.

DISCUSSION

In developing countries, HD has still been diagnosed late. In our series, the mean age at diagnosis was 6 years.

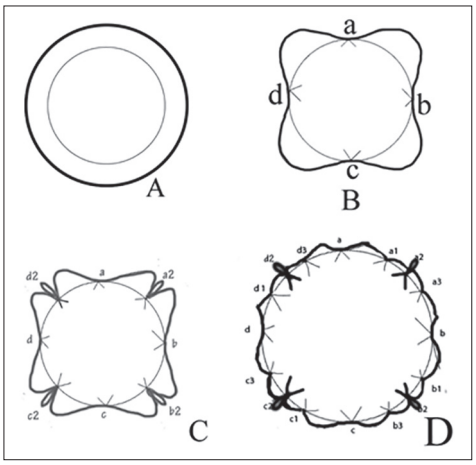

Figure 5: Plication procedure: (A) Incongruence between large dilated ganglionic colon and anal calibre. (B) Four initial quadrant sutures: a, b, c, d. (C) Four plicating sutures: a2, b2, c2, d2. (D) One to two stitches between each 2 previous sutures: a1, a3, b1, b3, c1, c3, d1, d3

The first reason for this delayed presentation included inadequate access to specialized centres and delayed referral. The second reason is that only 90% of all cases of aganglionosis produce clinical signs during the newborn period.[2] The situation is not different in many other countries, and the late presentation was attributed to illiteracy, parental ignorance and poverty.[3,4] In the present series, the incidence of late presentation is high (29 %) as compared to only 1-10 % in the developed world,[2,3,5] but still relatively low compared to other developing countries 72%.[6] Clinical features which point to diagnosis of HD include failure to evacuate meconium within 24 h of birth.[7] It is only reported in 9 of our patients but for 6 of them this data is forgotten by the family inspite of its high diagnosis value. The time of meconium evacuation is not reported systematically in the medical record after birth in our country. Chronic constipation dating back to the newborn period without encopresis is the characteristically presenting symptom of late diagnosed HD reported,[6] this feature is found in all our patients. Abdominal distension and palpation of large faecaloma are frequent features in, respectively, 10 and 7 patients of the present series. Malnutrition and anaemia which are important to late diagnosed HD as they require to improve the nutritional status and the haemoglobin level.[6,8] In the present series, 5 patients were malnourished and 1 with severe anaemia required transfusion. Approximately, 10% of patients with HD presented with HAEC.[9] Children with late diagnosed HD after neonatal period paradoxically resist to the development of HAEC because of the improved mucosal defenses.[10] However, in our series, 6 patients were admitted with HAEC may be because the diagnosis of HAEC was overestimated. However, it is better to err on the side of caution than to permit the child to become septic from untreated HAEC.[9] With a late diagnosed HD,

Table 3: postoperative results

Case n°	Perianal excoriation	Hirschsprung Associated entrocolitis	Anastomotic stricture	Soiling	Incontinence score	Constipation score	Follow-up duration (year)
1	YES	YES			25,5	26,5	6
2			YES		26	18,5	1
3					24,5	20	3
4	YES				24,5	22,5	1
5					24,5	22,5	2
6		YES		YES	22,5	16,5	2
7					24,5	20,5	1
8					24,5	20,5	1
9					24	23	4
10	YES	YES		YES	24,5	15,5	6
11		YES			24,5	21	4
12					24,5	22,5	1
13				YES	28	11	1
14			YES		—	—	1
15			YES		24,5	22,5	5

the extent of disease may explain the mild symptoms, which may contribute to a delayed diagnosis.[11] The meta-analysis of HD presented after childhood reported that the extent of the disease was in 79.6% to the rectum, 12.3% had the rectosigmoid disease, 0.8% extended to the descending colon, 0.4% extended to the transverse colon and 0.4% total colonic disease.[11] Other authors reported only rectosigmoid disease.[3,6,12] In the present series, 9 were confined to the rectosigmoid, 2 had disease extending to the descending colon, and 4 had the short disease. Manometry was performed in 6 patients; other authors adopted it in selected cases if there was a doubt in diagnosis.[6] Suction rectal biopsy is adequate to provide sufficient tissue in children aged bellow 3 years but for older children, a full-thickness biopsy is recommended.[9,10] In our practice, we used surgical rectal biopsy under general anaesthesia to confirm the diagnosis. Probiotics had a preventive role against HAEC by restoring bacterial equilibrium and stimulating secretion of immunoglobulin A.[10] In our practice, we use it during pre-operative period and after surgery. The pre-operative preparation was an important part of management. It helps for decompression, diagnosis, better operative procedure and less post-operative complications. Two ways are used for this preparation: Non-operative bowel preparation, using irrigation and/or laxatives for a mean period of 4 months.[12] In our experience, 12 patients were prepared only by rectal washout and the mean time of bowel preparation was 67 days. This long period of preparation required a good parental compliance. When non-operative management fails, colostomy or ileostomy can be used adequately to decompress the colon.[3,12,13] Unfortunately, severe colonic distension in the late diagnosed HD can increase the difficulty of maturing a loop colostomy and the colostomy related complications such as prolapse, bleeding, skin inflammation, dehiscence and incisional hernia.[3,8,14] We think as most of the patients had a rectosigmoid disease the position of the sigmoid loop colostomy risks to be in a non-functional colon. Ileostomy can lead to severe malnutrition in these already malnourished patients. We recommend a blowhole transverse colostomy which is a safe, quick and effective procedure that can help to decompress and facilitate colonic irrigation with less complications in those redundant and large dilated colons. We performed it without complications. Blowhole colostomy was first formally described by Turnbull *et al*. represents a minimally invasive form of colonic decompression. It was once used for the emergent management of toxic megacolon.[14] One stage EPT was reported successfully in other series[12] and can be a good option in developing countries providing quality care in one hospitalization and preventing the stigma of having a colostomy,[8] we performed it in 12 of our patients. The dissection was difficult at the beginning of the procedure in older patients. We think that the one stage endorectal pull-through is a safe procedure, reduce hospital stay and morbidity related to the colostomy. One stage transanal endorectal pull-through in the late diagnosed HD represent no risk for the sphincter mechanism, if we respect the rule of the dissection outside of the anus without retraction of the anus sphincter, and the hugely dilated colon tends to decompress after an efficient bowel management making the dissection outside the anus without retraction safer for the sphincter mechanism during OSEP. Further more, we must commence the dissection with the anastomosis 1-2 cm above the dentate line as recommended by De La Torre and Langer.[15] Multistage management may increase the overall morbidity.[3,8] To our knowledge, up till now two techniques have been reported to treat the anastomosis incongruence: Tapering[16] and step by step.[17] We described a simple technique by plication [Figure 5] using 16-24 stitches. We performed it in all our patients; we think it required fewer stitches than step by step and without tapering so less risk of leakage, fistula, anastomotic stricture and abscess. Patients with late diagnosed HD seem to be a higher risk of complications: Intraoperative bleeding, frequent bowel movement, temporary faecal incontinence with negative effect on mental health, anastomosis dehiscence, soiling and mortality due to associated enterocolitis.[3,4] Constipation and incontinence are the most important markers to assess the outcome of the patients. This assessment is achieved by the PICSS. Children who scored below their age-specific lower limit 95% CI PICSS scores were considered to have incomplete continence or constipation. At the telephone interview, all our patients are over 35 months, the incontinence score of our patients fall within the age-related 95% CI for some patients and superior to upper CI 95% limit for 3 patients. All our patients can be categorised as children with normal continence function. Constipation score of the patients 6, 10 and 13 was below the lower CI 95% limit and indicates the presence of constipation symptomatology only in those 3 patients. We found also the presence of soiling in the same 3 patients. Soiling is not secondary to incontinence but is a manifestation of overflow in those 3 constipated patients. These patients need botulinum injections to the internal sphincter.

CONCLUSION

Late diagnosed HD requires a challenging management by adequate bowel and general preparation. Rectal wash out is the best way to prepare the colon even it is a demanding

procedure, but when it becomes ineffective, the blowhole transverse colostomy can be an option. OTEPT is a safe option with less morbidity. Plication procedure we have described avoids anastomosis incongruence.

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 initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Nil.

Conflicts of interest

There are no conflicts of interest.

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