

Case Report

TOTAL COLONIC AGANGLIONOSIS: A RARE ENTITY IN CHILDREN

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ABSTRACT

Total colonic aganglionosis being an unusual form of Hirschsprung's disease is described in two children. Both the patients were male, one 8-year old and the other 12-year old. They presented with features of sub acute intestinal obstruction preceded by a past history of chronic constipation since birth and operative intervention for similar complaints few years back. A high index of suspicion prompted histopathological examination of colonic biopsies and the diagnosis of total colonic aganglionosis was confirmed. Two stage Martin's Duhamel retrorectal pull through resulted in satisfactory recovery. Total colonic aganglionosis represents a significant challenge for pediatric surgeons. Long-term results are suboptimal, complications are very common. The goal is to provide the patient with a good quality of life. Authors review their experience with this rare entity and the pertinent literature.

Key Words: *Total Colonic Aganglionosis, Intestinal Obstruction, Children*

INTRODUCTION

Total Colonic Aganglionosis also known as Zuelzer Wilson syndrome is an unusual form of Hirschsprung's disease. Total Colonic Aganglionosis constitutes only a small proportion of cases with Hirschsprung's disease and it usually presents in the first week of life owing to the severity of obstruction and gastro intestinal dysfunction (Lall *et al.*, 1999; Ikeda and Goto, 1986) but, still with reasons unknown, a few cases of total colonic aganglionosis may present in childhood. To the best of our knowledge only a handful of such cases have been reported in the English literature till date (Lall *et al.*, 1999; Caresky *et al.*, 1982). Moreover a high index of suspicion required for the diagnosis of this rare entity demands special recognition, with the aim to avoid an undue morbidity and mortality associated with its delayed diagnosis and the resulting complications. Thus rarity justifies reporting of the present cases with the aim to make physicians aware of clinical features and the management protocol of this unusual form of Hirschsprung's disease.

CASES

Case Number 1



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An 8 year old boy presented with abdominal pain, vomiting, constipation and progressive abdominal distension for the last 4 days. There was past history of chronic constipation present since birth for which the patient required persistent use of laxatives and enemas. The patient's attendant also gave history of previous surgical intervention done three years back for similar complaints when laparotomy was performed, but, the child didn't have amelioration of symptoms. The operative record was not available with the patient. On examination the patient appeared malnourished, anemic, and dehydrated with a pulse rate of 100/min, blood pressure of 110/70mm of Hg and a normal body temperature. Abdominal examination revealed a soft, non tender, distended abdomen with visible bowel loops and increased peristaltic activity on auscultation. Digital rectal examination suggested presence of soft fecal matter in the rectum.

Lab investigations suggested haemoglobin of 7gm% with normal total leukocyte counts, renal and liver function tests. Plain radiographs of abdomen revealed dilated bowel loops with multiple air fluid levels, thereby suggestive of intestinal obstruction. This precipitated surgery. Surgical exploration revealed dilated and thickened distal ileum with progressive dilatation towards the ileocecal junction. Terminal ileum, ileocecal junction and the colon was of normal caliber and after thorough surgical exploration no obvious cause of obstruction could be found. The presence of chronic constipation since birth with an absence of an obvious cause of intestinal obstruction during surgical exploration provided a clue to the diagnosis. Hence multiple seromuscular biopsies from colon and terminal ileum were taken and sent for histopathological examination along with rectal biopsy especially to look for features of Hirschprung's disease. Histopathological examination confirmed diagnosis of total colonic ananglionosis. A two stage Martin's modified Duhamel retrorectal pull through was performed with initial ileostomy as 1st stage procedure followed by definitive pull through after six months. Postoperative period remained uneventful and the patient recovered satisfactorily. The patient is presently doing well on follow-up for the last 3 years.

Case Number 2

A 12 year old boy presented with progressive abdominal distension for the last few days and chronic constipation since birth. There was past history of persistent use of laxatives and enemas and history of previous surgical intervention done two years back for sub acute intestinal obstruction. Records revealed that surgeon was not sure of the diagnosis, hence no mention about the intraoperative findings were made. Postoperatively the child didn't have relief from constipation. On examination the patient appeared thin built, malnourished, anemic, and dehydrated with a pulse rate of 110/min, blood pressure of 100/66mm of Hg. Abdominal examination revealed a midline incision with features consistent with sub acute intestinal obstruction with visible bowel loops.

Lab investigations suggested haemoglobin of 8gm%. Plain radiographs of abdomen revealed dilated bowel loops with multiple air fluid levels, thereby suggestive of intestinal obstruction. Barium enema showed uniformity of whole of the colon, absence of the haustrations and absence of the classical cone.

On exploration colon was collapsed and ileum was distended. After thorough surgical exploration no obvious cause of obstruction could be found. As we had become wiser from the previous case, multiple seromuscular biopsies from colon and terminal ileum were taken and sent for histopathological examination to look for Hirschprung's disease and 1st stage Mikulicz operation was performed. Histopathological examination confirmed diagnosis of total colonic ananglionosis. After six months Martin's modified Duhamel retrorectal pull through was performed. Postoperatively patient had an excellent recovery and is doing well.

DISCUSSION

Total colonic aganglionosis (TCA) with or without involvement of the small intestine is a rare form of Hirschsprung's disease (HD) and accounts for approximately 3% to 12% of all infants with HD, but its delayed presentation in childhood is extremely uncommon valve (Lall *et al.*, 1999; Ikeda and Goto, 1986; Basnet and Zheng, 2006; Moore, 2012; Bischoff *et al.*, 2011). Total colonic aganglionosis is defined as

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aganglioneosis extending from the anus to at least the ileocecal valve, but not >50 cm proximal to the ileocecal valve (Moore, 2012).

A review of cases with total colonic aganglioneosis by Ikeda and Goto suggest that 67.4% of total colonic aganglioneosis cases present within first month, 86.8% within first 3 months, 96.9% within the first year and none present beyond 13 months of age (Ikeda and Goto, 1986). According to another study 80% of patients are encountered in the neonatal period with delayed passage of meconium, and only a few patients presented in childhood and beyond (Basnet and Zheng, 2006). To our knowledge only few cases of total colonic aganglioneosis with delayed presentation in children and adults have been reported in English literature till date (Lall *et al.*, 1999; Crocker, 1991; Martin, 1972; Stone *et al.*, 1966; Myers *et al.*, 1966; Caresky *et al.*, 1982). But surprisingly this rare entity has been rarely discussed in Indian literature, thus demanding recognition of its typical clinical features and the management protocol.

It is not yet clear whether TCA merely represents a long form of HD or a different expression of the disease. There are many differences between TCA and other forms of HD. Clinically; TCA appears to represent a different spectrum of disease in terms of presentation and difficulties that may be experienced in diagnosis, suggesting a different pathophysiology from the more common forms of HD (Bischoff *et al.*, 2011).

The factors justifying the presentation of total colonic aganglioneosis in childhood remain unclear and are still an enigma for the investigators. There is also some evidence suggesting that instead of being purely congenital, it may represent certain different pathophysiologic mechanisms (Bischoff *et al.*, 2011). The passage of liquid ileal contents through entire aganglionic colon in small amounts explain the presentation of cases with total colonic aganglioneosis in infancy but the progression of disease into childhood remains unanswered (Moore, 2012).

The clinical presentation in the present cases is replica of typical features being described by most investigators in the available literature. Most of the cases present with features suggestive of sub acute intestinal obstruction and most of the cases have under gone repeated laparotomy for sub acute intestinal obstruction without permanent relief of symptoms (Lall *et al.*, 1999). Similarly as experienced in the present and reported cases of presence of chronic constipation since birth requiring persistent use of laxatives and anemia provide a clue to diagnosis (Lall *et al.*, 1999; Ikeda and Goto, 1986; Crocker, 1991). So a thorough clinical evaluation is mandatory in such cases especially to search for presence of symptoms of chronic constipation requiring persistent use of enemas and laxative and which remains an important diagnostic clue.

As experienced in the present and a few reported cases the intra operative findings and the results of radiological investigations including contrast enema may fail to suggest features of Hirschsprung's disease¹. So the diagnosis of total colonic aganglioneosis in such children remains a diagnostic challenge demanding awareness and a high index of suspicion based on a thorough clinical evaluation especially to search for presence of history of constipation since birth with an absence of amelioration of symptoms of subacute intestinal obstruction even after repeated laparotomies (Lall *et al.*, 1999).

Initial aim is histopathological confirmation of diagnosis followed by definitive treatment of Hirschsprung's disease. So a routine seromuscular colonic biopsy is routinely recommended in children presenting with repeated episodes of sub-acute intestinal obstruction especially in presence of a high index of suspicion. Frozen section examination if available remains a useful diagnostic modality for this purpose with its inherent advantages of prompt intraoperative diagnosis followed by definitive treatment at the same time which thereby prevents inadvertent resection of ganglionic distal ileal segment and hence preserves intestinal length (Lall *et al.*, 1999). But as experienced in the present case, in the absence of frozen section facility the management protocol in children with total colonic aganglioneosis remains a challenge for the treating physician. In such cases a routine histopathological confirmation of diagnosis by multiple seromuscular colonic biopsies followed by a definitive treatment of Hirschsprung's disease remains the treatment of choice. A routine stoma formation as initial surgical approach appears

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unjustified in such cases in view of extreme rarity of the disease. Similar surgical intervention resulted in a satisfactory recovery in the present case.

Thus we concluded that although rare, but total colonic aganglionosis should be suspected in children presenting with recurrent episodes of intestinal obstruction especially in presence of high index of suspicion based on presence of associated history of chronic constipation and a lack of amelioration of symptoms even after repeated laparotomies. The management in patients with total colonic aganglionosis represents a significant challenge for paediatric surgeons. Patients often undergo multiple complications in the form of ileostomy prolapse, strictures, rash, infections, fecal fistula, pullthrough of aganglionic bowel, destroyed anal canal and fecal incontinence and reoperations (Bischoff *et al.*, 2011). It's possible to provide the patient with a good quality of life and also by preventing complications by delaying ileostomy closure, having pathology expertise, and using meticulous surgical technique by starting the anastomosis above the dentate line to preserve the anal canal and sphincters.

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