Zonal adult Hirschsprung's disease

C G Fu, T Muto, T Masaki, H Nagawa

Abstract

Background—Hirschsprung's disease is a congenital disorder which is rare in adulthood. In typical cases the aganglionosis involves mainly the rectum or rectosigmoid colon and the lesion starts from the anal valve. Zonal segmental aganglionosis is a very rare type even in children.

Patient—A 54 year old women with zonal segmental aganglionosis had an aganglionic segment 18 cm in length located in the rectosigmoid colon with an 8 cm long normal appearing rectum and dilated proximal colon. Resection of the stenotic segment with end to end anastomosis was performed.

Conclusion—The functional result was excellent five years after the operation.

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Keywords: adult Hirschsprung's disease, constipation, megacolon.

Hirschsprung's disease is a congenital disorder which is rare in adulthood.^{1 2} In typical cases, the aganglionosis involves mainly the rectum or rectosigmoid colon and the lesion starts from the anal verge proximally for varying distances. The involved segment is narrowed and in a tonic state due to the lack of parasympathetic ganglion cells in both intramural and submucosal plexuses and the unopposed sympathetic activity.³⁻⁵

Zonal segmental aganglionosis, a very rare type even in children, in which there is a localised segment of colonic aganglionosis with the presence of myenteric ganglionic cells above and below the aganglionic segment was first described by Tiffin *et al*⁶ in 1940. Since then, although more cases have been reported,⁷⁻¹² the existence of this rare type of aganglionosis is still controversial.^{13 14}

We report an adult case with zonal colonic aganglionosis.

Case report

A 54 year old women had had life long refractory constipation. At the age of 9 years, a transverse colostomy was performed due to long lasting refractory constipation. This was closed at the age of 23 at the patient's request. Constipation recurred immediately after the operation. On admission, a barium enema study showed a persistent annular stenotic segment of the rectosigmoid colon 16 cm in length, 1.5 cm in luminal diameter, starting about 8 cm above the anal verge. The calibre of the distal rectum was normal. The proximal sigmoid colon was very dilated with fecalithes

inside (Fig 1). Fibrocolonoscopy showed normal mucosa in the stenotic segment and the distal rectum. Rectoanal reflexion examination disclosed that the fall in anal pressure was less than normal when a rectal balloon was inflated with 60 ml air and there was a stronger overshoot of the anal pressure after that than in normal patients. In defecography, barium in the distal rectum could be defecated, but with a longer time. Biopsy examination suggested that ganglion cells in the distal rectum were decreased in number whereas no ganglion cells could be found in the narrowed rectum. Acetyl cholinesterase staining was negative in a biopsy specimen of the narrowed part, which was taken by colonoscopy and was later considered as insufficient in depth. Unfortunately, staining was not performed on the resected specimen.

Routine laboratory evaluation, including a complete blood cell count, urine analysis, and blood chemistry studies, was normal.

Due to the extremely dilated proximal colon and difficulty in preparing the bowel, diverting transverse colostomy was performed at the first admission followed by definitive operation with closure of the colostomy nine months later. The narrowed segment of the rectum was resected down to the normal appearing rectum and anastomosis was performed about 5 cm above the anal verge. The resected specimen of

Figure 1: Narrowed segment of rectosigmoid colon with normal appearing distal rectum and dilated proximal colon shown by barium enema examination.

First Department
of Surgery,
University of Tokyo,
Tokyo, Japan
C G Fu
T Muto
T Masaki
H Nagawa

Department of
Colorectal Surgery,
Changhai Hospital,
Second Military
Medical University,
Shanghai, People's
Republic of China
C G Fu

Correspondence to:
Professor Tetsuichiro Muto,
First Department of Surgery,
University of Tokyo, 7-3-1
Hongo Bunkyo-ku, Tokyo,
Japan, 113.

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Figure 2: Resected specimen showing the narrowed segment, the proximal dilated colon, and the normal appearing distal rectum with two transitional zones (arrows).

Figure 3: Aganglionic zone with hypertrophied nerve trunks (haematoxylin and eosin originally $\times 100$).

Figure 4: Decreased distribution of ganglion cells (arrow) in the distal normal appearing rectum (haematoxylin and eosin originally $\times 100$).

the colon was 20 cm in length after fixation. Figure 2 shows two transitional zones, which divide the rectosigmoid colon into three parts: the proximal dilated part (Fig 2A), the stenotic

part (Fig 2B), and the distal normal appearing part (Fig 2C). The stenotic segment (Fig 2 between the two arrows) was 12 cm in length and 4 cm in width; the width of the proximal colon was 10.5 cm and the distal rectum 6 cm. The colour of the narrowed and the distal rectum was normal, whereas there was melanosis in the proximal colon. Ganglion cells were not found in the submucosal or intramural layers in the narrowed part of the colon histopathologically, and nerve trunks in this part were both hypertrophied (Fig 3) and increased in number. The number of ganglions or ganglion cells in each ganglion in the distal normal appearing rectum were less than those in the proximal dilated colon (Fig 4 and Fig 5). Postoperative recovery was uneventful and the patient enjoyed defecation once a day after operation, with differentiating gas and stool. No incontinence was encountered and no laxatives was needed at least until five years after the operation.

Discussion

Hirschsprung's disease is a congenital disease which occurs in about one in 5000 births. Ninety four per cent are diagnosed before the age of 5 years. In rare cases, the patients may be undiagnosed until adulthood.^{1 15}

In typical Hirschsprung's disease, the aganglionosis starts from the anal valve, spreads proximally, and involves only the rectum and sigmoid colon. This clinical feature concurs with embryological studies, which showed that the myenteric plexus arose from neuroblast migration into the alimentary tract in a cranial to caudal direction during the fifth to 12th week of gestation.¹⁶ Thus when abnormalities occur, the distal part of the bowel is most likely to be involved. This results in the loss of relax ability of the involved rectum and absence of rectoanal reflexion. In less than 10% of the patients, aganglionosis may occur more proximally and in some cases even the ascending colon or small intestine may be involved.^{1 4}

Zonal aganglionosis 'in which there is an area of colonic aganglionosis with the presence of myenteric ganglion cells above and below the aganglionic segment' is a very rare type of this disease.⁷⁻¹² Since 1940, when Tiffin *et al* first reported a case with zonal aganglionosis, only 15 cases have been reported¹² and among them only two were adults. The reason for this specific type is unclear. Kadair *et al* speculated that it might be the result of a 'skip' of neuroblast migration and distribution in the embryonic development period.⁹ In most reported cases, the distribution of the ganglion cells in the distal part of the colon or rectum was normal. However, in this patient, the number of ganglion cells in the distal rectum was decreased compared with that in the proximal colon and no ganglion cells could be found in the narrowed segment. This might be explained by the abnormality in embryological development.

In adults, the diagnosis of Hirschsprung's disease should be made with care, as patients with idiopathic megacolon may also be

Figure 5: Normal distribution of ganglion cells (arrows pointed) in the proximal part of the sigmoid colon (haematoxylin and eosin originally $\times 100$).

constipated and have a dilated colon. Furthermore, some acquired problems may also result in disappearance of the ganglion cells in both submucosal and intramural layers and lead to persistent contraction of the involved colon and megacolon proximally, such as in Chagas' disease.¹⁷ Thus, as in this case, the patient should not only have a narrowed segment of the colon or rectum with dilated proximal colon, but also a lifelong history of refractory constipation for making the diagnosis. Laboratory examinations, such as rectoanal reflexion, defecography, and acetyl cholinesterase staining should also be considered if the diagnosis is suspected, and the results should support the diagnosis. It should also be kept in mind that there might be some exceptions. As in this case, although the results of rectoanal reflexion and defecography were not normal, there was rectoanal reflexion and the patient could defecate the barium in the distal rectum, but with a longer time. Also acetyl cholinesterase staining was negative in the biopsy specimen (later it was considered that the biopsy was not deep enough). So, the final diagnosis should be made on general

consideration of the history, laboratory examination, and histopathological examination of the stenotic segment of the colon or rectum. Doctors who treated this patient when she was 9 and 23 had suspected the diagnosis of Hirschsprung's disease, when they performed or closed the transverse colostomy, but unfortunately they were confused by the atypical clinical features and delayed the diagnosis. Otherwise, the patient would not have suffered from continuous constipation until the age of 54 years.

- 1 Ikeda K, Goto S. Diagnosis and treatment of Hirschsprung's disease in Japan. An analysis of 1628 patients. *Ann Surg* 1984; **199**: 400–5.
- 2 Fairgrieve J. Hirschsprung's disease in the adult. *Br J Surg* 1963; **50**: 506–14.
- 3 Bentley JF. Some new observations on megacolon in infancy and childhood with special reference to the management of megasigmoid and megarectum. *Dis Colon Rectum* 1964; **7**: 462–70.
- 4 Ikeda K, Goto S. Total colonic aganglionosis with or without small bowel involvement. An analysis of 137 patients. *J Pediatr Surg* 1986; **21**: 319–22.
- 5 Venugopal S, Mancor K, Shandling B. The validity of rectal biopsy in relation to morphology and distribution of ganglion cells. *J Pediatr Surg* 1981; **16**: 433–7.
- 6 Tiffin MD, Chandler LR, Faber HK. Localized absence of the ganglion cells of the myenteric plexus in congenital megacolon. *Am J Dis Child* 1940; **59**: 1071–82.
- 7 Swenson O, Rheinlander HF, Diamond I. Hirschsprung's disease: a new concept of the etiology. *N Engl J Med* 1949; **241**: 551–6.
- 8 Sprinz H, Cohen A, Heaton LD. Hirschsprung's disease with skip area. *Ann Surg* 1961; **153**: 143–8.
- 9 Kadair RG, Sims JE, Critchfield CF. Zonal colonic hypoganglionosis. *JAMA* 1977; **238**: 1838–40.
- 10 de-Chadarevian JP, Slim M, Akel S. Double zonal aganglionosis in long segment Hirschsprung's disease with a "skip area" in transverse colon. *J Pediatr Surg* 1982; **17**: 195–7.
- 11 McCreedy RA, Beart RW Jr. Adult Hirschsprung's disease: results of surgical treatment at Mayo Clinic. *Dis Colon Rectum* 1980; **23**: 401–7.
- 12 Moriya H, Nakazaki H, Yokoyama S, Tajima T, Mitomi T, Satou S. A case of segmental aganglionosis localized descending colon. *Jpn J Gastroenterol* 1996; **93**: 39–44. (In Japanese).
- 13 Yunis E, Sieber WK, Akers DR. Does zonal aganglionosis really exist? Report of a rare variety of Hirschsprung's disease and review of the literature. *Pediatr Pathol* 1983; **1**: 33–49.
- 14 Wheatley MJ, Wesley JR, Coran AG, Polley TZ Jr. Hirschsprung's disease adolescence and adults. *Dis Colon Rectum* 1990; **33**: 622–9.
- 15 Powell RW. Hirschsprung's disease in adolescents: misadventures in diagnosis and management. *Am Surg* 1989; **55**: 212–8.
- 16 Okamoto E, Ueda T. Embryogenesis of intramural ganglia of the gut and its relation to Hirschsprung's disease. *J Pediatr Surg* 1967; **2**: 437–43.
- 17 Todd IP, Porter NH, Morson BC, Smith B, Friedmann CA, Neal RA. Chagas' disease of the colon and rectum. *Gut* 1969; **10**: 1009–14.