

Hirschsprung's disease and idiopathic megacolon in adults and adolescents

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SUMMARY The distinction between Hirschsprung's disease and idiopathic megacolon in childhood dates from the classic clinical, radiological, and histological studies of Bodian, Stephens, and Ward.¹ This article describes clinical experience over 15 years of 94 patients in whom megacolon of these two types was recognised for the first time after the age of 10, to illustrate the problems of diagnosis and treatment in later years.

Just as it is now recognised that patients with the clinical characteristics of Hirschsprung's disease may have one of several abnormalities of the myenteric plexus, including not only absence of ganglion cells, but also patchy or zonal loss,^{2,3} abnormal neurones⁴ or neuronal dysplasia,^{3,5,6} so idiopathic megacolon may also be a heterogeneous group of cases. This paper suggests on clinical grounds that those patients with idiopathic megacolon whose symptoms start in childhood differ from those whose symptoms develop in later years.

In the years 1968 to 1983, 134 patients over the age of 10 years with Hirschsprung's disease or idiopathic megacolon were investigated and treated as inpatients at St Mark's Hospital. Of the 51 patients with Hirschsprung's disease, the 29 diagnosed and treated for the first time after the age of 10 years are reported here; the remainder (22) had Hirschsprung's disease diagnosed previously, usually in early childhood. Megacolon of the non-Hirschsprung's type, defined by histological and physiological studies, was seen in 65 patients; patients with an apparent cause for the megacolon, such as previously treated anal atresia, were excluded as were 16 patients with incomplete diagnostic data.

Follow up information was obtained in 82% of the patients, either from notes made at outpatient and inpatient consultations or from a postal questionnaire where necessary.

Methods

The diagnosis of megacolon was made by barium enema. The distinction between Hirschsprung's disease and the non-Hirschsprung's type was made by a combination of histological examination of

biopsies or operation specimens and the presence or absence of the rectosphincteric reflex.

BARIUM ENEMA EXAMINATION

(a) *Hirschsprung's disease* (29)

A previous study⁷ based on radiographs from 20 patients in this series compared with a control group has shown that megacolon can be defined by measurements of bowel width and that such a definition corresponds well with the opinion of radiologists. Where possible a definition of megacolon as exceeding 6.5 cm in width at the rectosigmoid junction has been taken but in many cases the radiological opinion had to be accepted as films were no longer available.

All 29 patients had x-rays performed or reviewed at St Mark's Hospital. The barium enema was suggestive of Hirschsprung's disease in 22, reported as megacolon without an obvious short-segment in six and was reported as being normal in one. The upper limit of the distal narrowed segment was in the rectosigmoid in 20 (Fig. 1) and in the descending colon in two.

(b) *Non-Hirschsprung's megacolon* (65)

All 65 patients had a barium enema examination. Megarectum was reported in 31, megarectum and megasigmoid in 10 (Fig. 2), and in 24 the upper limit of the widened bowel was not reported.

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Fig. 1 Barium enema showing the typical distal narrow segment of Hirschsprung's disease.

HISTOLOGY

(a) Hirschsprung's disease (29)

Rectal biopsy of the full thickness type with conventional histological staining revealed typical appearances of Hirschsprung's disease in 24 patients and in three the findings were suggestive but not diagnostic. Two of the latter had confirmation of the diagnosis on examination of an operative specimen and one on the basis of an absent rectosphincteric reflex. Two patients did not have a biopsy done before surgery but in both cases the diagnosis was confirmed on examination of the operative specimen.

(b) Non-Hirschsprung's megacolon (65)

Fifty four patients had a rectal biopsy which was normal in 52 and equivocal in two.

RECTOSPHERIC REFLEX

(a) Hirschsprung's disease (29)

Nineteen patients were tested pre-operatively for the presence of a rectosphincteric reflex. The reflex

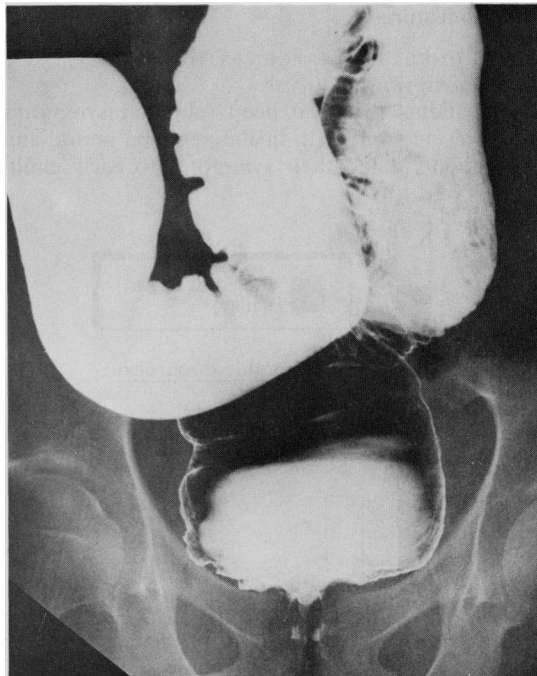


Fig. 2 Barium enema appearance of megarectum and megasigmoid.

was absent in 18 and reported as present in one patient who had typical features of Hirschsprung's disease on a subsequent rectal biopsy.

(b) Non-Hirschsprung's megacolon (65)

In the 41 patients tested, the reflex was present in 34 and absent in seven. Those patients in whom the reflex was absent all had normal histology on biopsy.

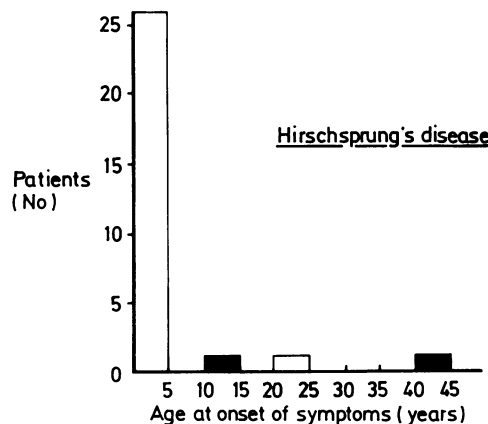


Fig. 3 Age at onset of symptoms in Hirschsprung's disease presenting after the age of 10 years.

Clinical features

AGE AT ONSET AND PRESENTATION (Fig. 3)

(a) Hirschsprung's disease

Most patients (21) had been told that symptoms were present from birth or the neonatal period and five patients dated their symptoms to early child-

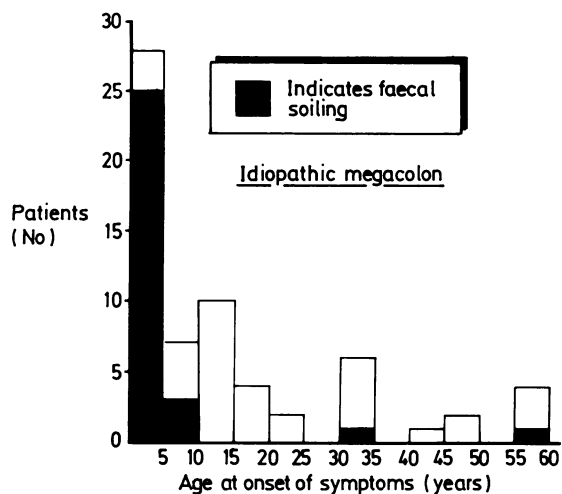


Fig. 4 Age at onset of symptoms in idiopathic megacolon presenting after the age of 10 years.

hood (below the age of five years). Two patients first developed symptoms in adolescent years before the age of 20 and one man of 73 reported that his symptoms began at the age of 40. One patient first developed symptoms during pregnancy and in two others symptoms were worse at this time. Mean age at presentation was 26 years (range 11–73 years).

(b) Non-Hirschsprung's megacolon (Fig. 4)

It was apparent that a large proportion of patients with megacolon of the non-Hirschsprung's type had the onset of symptoms in early childhood and these cases commonly had symptoms of faecal soiling and were impacted on rectal examination. Other patients first noted symptoms at various ages throughout later life and in general did not have faecal soiling or gross rectal impaction. For the purpose of analysis, patients with non-Hirschsprung's megacolon were thus divided into two groups, based upon the age at which symptoms developed, arbitrarily defined as less than or equal to 10 years (childhood megacolon) and greater than 10 years (adult megacolon).

The group of 35 patients with childhood onset first developed symptoms at a mean age of 3.4 years (range from birth – 10 years) and presented at a mean age of 16 years (range 10–27 years). The group of 30 patients with later development of symptoms

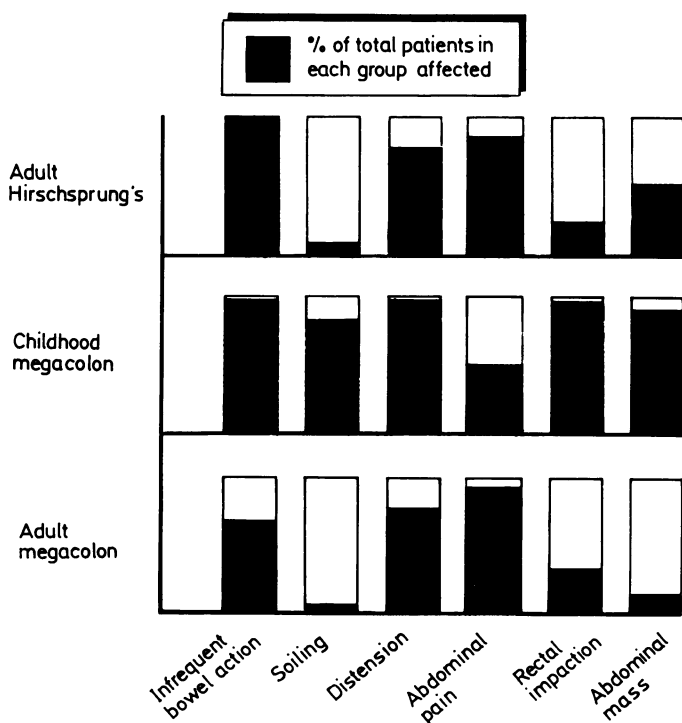


Fig. 5 Comparison of the symptoms and signs at presentation after the age of 10 years in Hirschsprung's disease, idiopathic megacolon with onset of symptoms in childhood, and idiopathic megacolon with onset of symptoms in adolescence or adult life.

had a mean age of onset of 29 years (range 11–59 years) and presented at a mean age of 38 years (range 15–68 years).

SEX

The patients with Hirschsprung's disease comprised 19 men and 10 women. There were 23 men and 12 women with idiopathic megacolon whose symptoms began in childhood and 17 men and 13 women who first developed symptoms after the age of 10 years.

HISTORY, SYMPTOMS AND SIGNS AT PRESENTATION (Fig. 5)

(a) Hirschsprung's disease

All patients in the Hirschsprung's group reported

constipation as the predominant symptom, and most evacuated the bowel only with enemas. Abdominal pain was also a common symptom (24 patients) as was abdominal swelling (22 patients). Only two patients reported faecal soiling, this developed in a girl of 14 after a manual dilatation of the anus for constipation and in a man with a 30 year history of constipation just before his first consultation at the age of 73. An abdominal mass was palpable in 16 patients and seven had stool in the rectum on first examination. In most cases the rectum was empty, faeces being encountered at a higher level with the sigmoidoscope. Three patients gave a family history of Hirschsprung's, two in siblings and one with an affected child.



Fig. 6 *Gastrograffin (water soluble) contrast enema showing faecal impaction in idiopathic megacolon.*

(b) Idiopathic megacolon onset in childhood

Severe constipation was the major symptom in 34 of 35 patients in this group, one patient reported diarrhoea (possibly overflow soiling associated with rectal impaction). Faecal soiling had been troublesome in 28, 34 had symptoms of abdominal swelling and 19 reported abdominal pain or discomfort. An abdominal mass was palpable in 33 and 34 had rectal impaction at the first visit (Fig. 6). The onset of symptoms was related to a non-specific, possibly viral illness in two, an anal fissure in one and the use of anti-diarrhoeal drugs in one. Six patients were thought to be socially or psychologically disturbed and two were mentally retarded, but in the remainder no precipitating event or underlying cause was recorded.

(c) Idiopathic megacolon onset in adult life

Constipation was the major symptom in 19, but seven complained predominantly of diarrhoea and four had alternating diarrhoea and constipation. Abdominal pain was a symptom in 28 patients and 23 reported abdominal swelling. In contrast with the group with onset in childhood, only two patients in this group had faecal soiling and incontinence and this was only associated with attacks of diarrhoea; neither had rectal impaction on initial examination. Faecal impaction was only recorded in 10 of these patients and in some the rectum was dilated and empty, the sigmoidoscope 'falling in' to 25 cm. Only four cases had a palpable abdominal mass.

Eleven patients in this group had had previous surgical treatment, in two patients rectopexy for prolapse, in six a sigmoid colectomy, in four a resection with caecorectal anastomosis and in one a left hemicolectomy. Two patients had grand mal seizures and were on long term anti-convulsants and two had psychological disorders requiring treatment with major tranquillisers. Three patients were thought to have low intelligence although formal testing was not done. No precipitating event or predisposing factor was identified in the remainder.

Results**HIRSCHSPRUNG'S DISEASE**

All 29 patients in this group (19 men, 10 women) were treated surgically, 20 with a Duhamel operation, eight with resection and Soave coloanal anastomosis and one with internal sphincterotomy.

(i) Duhamel operation (20 patients)

Follow up of 17 patients was satisfactory over a mean of 4.0 years (range 3–16 years) and a mean of 4.5 visits. Of the 17 patients, 16 reported good results with regular bowel function (usually several

times a day), although occasionally some took laxatives. One patient whose postoperative course was complicated by a pelvic abscess had persistent constipation and soiling. Two other patients also experienced mild intermittent soiling. One patient complained of pelvic pain and dyspareunia and required trimming of the rectal septum.

(ii) Resection and coloanal anastomosis

Of eight patients in this group, seven were followed regularly (or responded to the questionnaire) over a mean of 2.8 years (range 1–7 years) with a mean of four visits. Seven reported regular bowel function, two with mild faecal soiling, while one remained constipated and took laxatives.

(iii) Internal sphincterotomy

The single patient treated with internal sphincterotomy remained well but still required osmotic laxatives and bulk forming agents three years after operation to maintain regular bowel actions.

IDIOPATHIC MEGACOLON ONSET IN CHILDHOOD

In this group of 35, 33 were followed for a mean of three years (range 1 to 15 years) with a mean of six visits.

(i) Medical treatment

Of the 26 patients treated medically, usually with osmotic laxatives and often with suppositories or enemas, 18 had a successful outcome with regular bowel actions and no soiling as long as they continued medication. A further five remained well while taking laxatives but when treatment was stopped developed recurrent constipation and impaction, and then often needed admission to hospital. Three patients had a poor result being unable to tolerate or adjust the dose of laxatives because of low intelligence, poor compliance, or side effects.

(ii) Surgical treatment

Nine cases were treated surgically, because of failure of medical treatment. Of four treated by colectomy and caecorectal anastomosis, two improved, one remained constipated and one did not respond to the questionnaire. One patient had a Soave coloanal anastomosis with poor results and required a colostomy. Of three who had a Duhamel procedure, two had regular bowel actions, (one with minor faecal soiling) and one did not respond to the questionnaire.

Five of nine surgical specimens showed a thick, hypertrophic bowel wall, one was thinned and three were reported as being of normal thickness.

IDIOPATHIC MEGACOLON ONSET IN ADULT LIFE

This group comprised 30 patients, 27 of whom were followed up for a mean of five years (range one–13 years) with a mean of six visits.

(i) Medical treatment

Eight received medical treatment alone, with laxatives, enemas and suppositories and four did well with regular bowel motions. One patient had a poor result with persistent symptoms and three did not respond to the questionnaire.

(ii) Surgical treatment

Surgical treatment was undertaken for 22 patients after failure of medical treatment. Of 10 treated by colectomy and caecorectal anastomosis, results were good in seven, with regular, often frequent bowel actions, without soiling, but three had persistent constipation and pain. Of six treated by colectomy and ileorectal anastomosis, four were improved with regular bowel actions, one remained constipated requiring laxatives and one did not reply to the questionnaire. Two patients were found to have developed megaileum after ileorectal anastomosis. Four patients had a partial colectomy with a good result in two and poor result in the other two because of persistent constipation. Five patients developed early or late adhesive obstruction after resection.

Examination of the surgical specimen revealed a thinned colonic wall in 12 and in six of these, examination by Smith's technique of silver staining, showed a disorder of the myenteric plexus. The remaining 10 specimens appeared normal on conventional histological examination but were not examined with Smith's technique.

Discussion

Hirschsprung's disease is diagnosed less often in adult life than idiopathic megacolon.⁸ The 29 patients with Hirschsprung's disease described in this paper were all diagnosed for the first time in adolescence or adult life. Although there are many papers describing Hirschsprung's disease presenting in childhood, there are few series of patients diagnosed after the age of 10 years, though first diagnosis in later years is well recognised.^{9–13} Although most patients gave a history of severe constipation, often needing repeated enemas since birth, in two patients symptoms were first noted in adolescence, and in one at the age of 40. In childhood there is a marked male predominance but in this series almost one third of the patients were female.

The term idiopathic (non-Hirschsprung's) megacolon has been used in this paper to describe a condition in which the diameter of the rectum and/or colon demonstrated by barium enema is increased¹⁴ but there is no evidence of an aganglionic segment. Measurements of radiographs from 20 of the 65 patients included in this paper for whom x-rays were available have shown that in every case the diameter of the distal bowel fell outside the normal range⁷ confirming the radiologist's opinion. The radiographs of the remaining 45 patients have been regarded by the radiologists as characteristic of megacolon but were not available for measurement.

The clinical analysis of the 65 patients with idiopathic megacolon suggests that two subgroups can be distinguished, one a group of patients whose symptoms began in childhood and the other group those who developed symptoms after the age of 10 years, often as adults. Among the former, faecal soiling with rectal impaction of stool was almost universal, but among the latter it was rare. A further important difference was that medical treatment was successful in the majority of patients with early onset of symptoms, whereas it was unsuccessful in the majority of those with onset in later life for whom surgical treatment was often needed. The limited anatomical data in six of the latter patients whose colonic myenteric plexus was studied by the silver staining technique^{15 16} showed an abnormality in every case. It is tempting to regard those with onset in childhood as the small proportion of children with rectal inertia^{17 18} whose trouble did not resolve before the age of 10, and those with later onset as having an acquired disorder of the myenteric plexus.

The distinction between Hirschsprung's disease and idiopathic megacolon can be difficult. A patient with a history of faecal soiling since childhood is most likely to have idiopathic megacolon, yet soiling does occur in a few children with short segment Hirschsprung's disease¹³ and in this series occurred in two of 29 patients, in one after anal dilatation and in the other over the age of 70 years. Abdominal pain, distension and a palpable abdominal mass are common to both conditions. Hirschsprung's disease cannot be excluded by absence of a narrow distal segment on barium enema.¹⁹ The recto-anal distension reflex can rarely, as in this series, be reported as present in Hirschsprung's disease.^{20 21} It has been suggested that in such cases the balloon used to distend the rectum may also dilate the anal canal or on distension it may pull the anal pressure probe upwards into the rectum.²² Conversely, the reflex may be absent in non-Hirschsprung's megacolon because distension of a balloon in a capacious

rectum may not act as a sufficient stimulus, or because the internal sphincter is already fully relaxed because of rectal impaction with stool or has been stretched by therapeutic manoeuvres such as manual evacuation of the rectum. Whenever there is doubt about the diagnosis, or the rectosphincteric reflex is absent, biopsy is essential.

The treatment of Hirschsprung's disease is surgical. It is noteworthy that only one of 29 patients was treated by extended internal sphincterotomy, though this operation has been recommended especially for children with short segment disease.²³ Successful results were obtained both with the Duhamel procedure and with resection as a Soave sleeve coloanal anastomosis, though the former is preferred.^{10 24}

Idiopathic megacolon may respond to either medical or surgical treatment. The majority of patients with onset of symptoms in childhood were managed successfully with medical measures. It must be emphasised that such patients tended to redevelop rectal impaction if laxatives were stopped and thus such patients must be advised to continue laxatives indefinitely. Unfortunately some patients are advised to stop treatment by their doctors who do not understand the nature of their problem. If rectal impaction reoccurs it can be difficult for the patient to empty the rectum even with enemas or washouts and manual removal of faeces from the rectum may be needed. In this group of patients with childhood onset, it is those patients who cannot comply with long term medical treatment who tend to need surgical treatment.

Patients who develop megacolon in adult life do not always suffer from constipation. Some complain mainly of abdominal pain and swelling with an irregular bowel habit, and sometimes episodes of diarrhoea. Among those who develop symptoms associated with a megacolon in adult life medical treatment is often unsatisfactory. The patients are greatly troubled by abdominal pain and distension, and laxatives for the treatment of constipation may be ineffective or produce uncontrollable diarrhoea. The majority of such patients in this series were offered surgical treatment for this reason. The results were variable but about two-thirds were helped by colectomy and ileorectal or caecorectal anastomosis.

The distinction between idiopathic megacolon of early and later onset suggested here on clinical grounds needs further investigation by physiological and anatomical methods. In particular, information is needed about the thickness of the muscle wall²⁵ and the integrity of the myenteric plexus in patients who develop symptoms for the first time at different ages.

References

- 1 Bodian M, Stephens FD, Ward BCH. Hirschsprung's disease and idiopathic megacolon. *Lancet* 1949; **1**: 6-11.
- 2 MacMahon RA, Moore CCM, Cussen LJ. Hirschsprung-like syndromes in patients with normal ganglion cells on suction rectal biopsy. *J Pediatr Surg* 1981; **16**: 835-9.
- 3 Nixon HH, Lake B. "Not Hirschsprung's disease" - rare conditions with some similarities. *S Afr J Surg* 1982; **20**: 97-104.
- 4 Emery JL. Colonic retention syndrome (megacolon) associated with immaturity of intestinal intramural plexus. *Proc R Soc Med* 1973; **66**: 222-3.
- 5 Meier-Ruge W. Hirschsprung's disease: its aetiology, pathogenesis and differential diagnosis. *Curr Top Pathol* 1974; **59**: 131-79.
- 6 Howard ER, Garrett JR, Kidd A. Constipation and congenital disorders of the myenteric plexus. *J R Soc Med* 1984; **77**: suppl 3: 13-19.
- 7 Preston DM, Lennard-Jones JE, Thomas BM. Towards a radiologic definition of idiopathic megacolon. *Gastrointest Radiol* 1985; **10**: 167-9.
- 8 Jennings PJ. Megarectum and megacolon in adolescents and young adults: results of treatment at St Mark's Hospital. *Proc R Soc Med* 1967; **60**: 7-8.
- 9 Tobon F, Schuster MM. Megacolon: special diagnostic and therapeutic features. *Hopkins Med J* 1974; **135**: 91-105.
- 10 Todd IP. Adult Hirschsprung's disease. *Br J Surg* 1977; **64**: 311-12.
- 11 Orr JD, Scobie WG. Presentation and incidence of Hirschsprung's disease. *Br Med J* 1983; **287**: 1671.
- 12 Rich AJ, Lennard TWJ, Wilsdon JB. Hirschsprung's disease as a cause of chronic constipation in the elderly. *Br Med J* 1983; **287**: 1777-8.
- 13 Doig CM. Childhood constipation and late-presenting Hirschsprung's disease. *J R Soc Med* 1984; **77**: suppl 3: 3-5.
- 14 Lane RHS, Todd IP. Idiopathic megacolon: a review of 42 cases. *Br J Surg* 1977; **64**: 305-10.
- 15 Smith B. *The neuropathology of the alimentary tract*. London: Arnold, 1972.
- 16 Smith B, Grace RH, Todd IP. Organic constipation in adults. *Br J Surg* 1977; **64**: 313-14.
- 17 Callaghan RP, Nixon HH. Megarectum: physiological observations. *Arch Dis Childh* 1964; **39**: 153-7.
- 18 Nixon HH. Megarectum in the older child. *Proc R Soc Med* 1967; **60**: 3-6.
- 19 Patriquin H, Martelli H, Devroede G. Barium enema in chronic constipation: is it meaningful? *Gastroenterology* 1978; **75**: 619-22.
- 20 Aaronson I, Nixon HH. A clinical evaluation of anorectal pressure studies in the diagnosis of Hirschsprung's disease. *Gut* 1972; **13**: 138-46.
- 21 Faverdin C, Dornic C, Arhan P, Devroede G, Jehannin B, Revillon Y, Pellerin D. Quantitative

- analysis of anorectal pressures in Hirschsprung's disease. *Dis Colon Rectum* 1981; **24**: 422-7.
- 22 Nagasaki A, Ikeda K, Suita S, Sumitomo K. Induction of the rectoanal reflex by electric stimulation. *Dis Colon Rectum* 1984; **27**: 598-601.
- 23 Udassin R, Nissan S, Lernau O, Hod G. The mild form of Hirschsprung's disease (short segment): fourteen years experience in diagnosis and treatment. *Ann Surg* 1981; **6**: 767-70.
- 24 McCreedy RA, Beart Jr RW. Adult Hirschsprung's disease: results of surgical treatment at Mayo clinic. *Dis Colon Rectum* 1980; **23**: 401-7.
- 25 Todd IP. Some aspects of adult megacolon. *Proc R Soc Med* 1971; **64**: 561-5.