Abstract: We report a 42-year-old male, a case of megacolon from adult hypoganglionosis. The patient suffered from intractable constipation since childhood and presented with abdominal distension. He also had surgery for sigmoid volvulus in the past. Barium enema and computed tomography revealed markedly dilated large bowel with a transition zone. A full thickness biopsy of the rectum showed a reduction in ganglion cells, thereby giving a diagnosis of adult hypoganglionosis of the colon or pseudo-Hirschsprung's disease.

Keyword: hypoganglionosis, aganglionosis, colon, transition zone

INTRODUCTION

Only a few cases of intestinal hypoganglionosis have been reported in literature, in view of its diagnostic difficulty. Adult hypoganglionosis is a rare innervation disorder of the intestine. The spectrum of neuronal disorders of the large bowel include aganglionosis or adult Hirschsprung's disease, hypoganglionosis and intestinal neuronal dysplasia which together are known as dysganglionoses (1). Pseudo-Hirschsprung's disease is a collective term used for disorders presenting with a clinical picture akin to Hirschsprung's disease. These include hypoganglionosis (decline in the number of intramural ganglion cells), intestinal neuronal dysplasia, immature enteric intramural neuronal cells, chronic idiopathic intestinal pseudo-obstruction and megacystis-microcolon-intestinal hypoperistalsis syndrome (2). Most of the cases of dysganglionoses are diagnosed in childhood with 94% being diagnosed before the age of 5. Few, with milder symptoms may go undiagnosed until adulthood. These patients present with refractory constipation which is manageable with bulk formers, enemas and other cathartic agents. Occasionally they may also present with severe symptoms like obstruction, perforation and volvulus. Hypoganglionosis (HG) as opposed to Hirschsprung’s disease (HD) is characterised by the presence of few ganglion cells and decreased number of parasympathetic nerves in the intestinal wall. The exact cause of adult hypoganglionosis is not known. However some authors have proposed an acquired etiology like circulatory disturbance, inflammatory, infectious cause (3,4). Pathological studies have distinguished two types of hypoganglionosis: Type 1 (focal type) has a focally narrowed transitional zone with few ganglion cells resulting in functional bowel obstruction and Type 2 (diffuse type) do not have a transition zone, but show diffuse reduction in ganglion cells along the entire colon (5). Accurate diagnosis of these innervation disorders from other causes of constipation allows definitive treatment, as these conditions are treatable with surgery (6). The primary imaging tools used for evaluation are barium enema and computed tomography (CT). The final diagnosis is based on histopathological evaluation of full thickness biopsy of the intestinal wall. However, some of the imaging features are more suggestive of HD than HG. These being, disease localised to the recto-sigmoid region and a higher transition zone ratio, i.e. the proximal bowel tends to be more dilated in HD(1). Hypoganglionosis on the other hand has varied forms – isolated HG, localized HG, disseminated HG, and HG combined with either HD or intestinal neuronal dysplasia. The treatment options depend on the length of segment involved with total colectomy being preferred for the disseminated form.

CASE REPORT

A 42-year-old man presented to our hospital with abdominal distension and intermittent pain for 3 months. This was associated with inability to pass stools or flatus for 3-4 days which would resolve spontaneously with passage of large amounts of stool. He did not have associated vomiting, melaena or hematemesis. Our patient also had a history of chronic constipation that was not evaluated and has been treated with laxatives and prokinetics. He had a prior history of sigmoid volvulus for which a resection and anastomosis was done. His family history and medical history were non-contributory. Physical examination revealed grossly distended abdomen with visible intestinal peristalsis. A digital rectal examination was normal. Initial laboratory test results revealed low haemoglobin and serum transferrin levels.

An Initiative of The Tamil Nadu Dr. M.G.R. Medical University
University Journal of Medicine and Medical Specialities
Fig 2. CT sections showing a) dilated ascending, transverse and descending colon. Computed tomography (CT) scan showed gross distension of sigmoid colon, descending, transverse and distal ascending colon measuring up to a maximum calibre of 12cm. The rectum was normal in calibre; with the transition zone being at the recto-sigmoid region, with no associated wall thickening (Fig.2).

Fig 3
Barium enema showing dilated sigmoid and descending colon with smooth narrowing at the rectosigmoid (transition zone). The proximal ascending colon is normal. Barium enema again showed a massively distended sigmoid colon, descending, transverse and ascending colon. A transition zone at the recto-sigmoid, measuring ~3.2cm was noted. The mucosal lining was smooth (Fig.3). He was planned for a loop ileostomy/transverse colostomy after adequate nutritional build up. Intraoperatively the distal transverse colon was distended with few flimsy adhesions between the bowel loops and peritoneum. Hence, subsequently a laparotomy, diversion loop transverse colostomy and full thickness rectal biopsy was done. Post-operatively our patient improved and remained well. The histopathological examination revealed hypertrophic nerve bundles and few ganglion cells, suggesting hypoganglionosis. He was discharged on a high protein diet, syrup magnesium sulphate and advised on stoma care. At the time of discharge, the stoma was functioning well and he was tolerating a normal diet.

DISCUSSION

Innervation disorders of the intestine are less often diagnosed in adults. In view of its rarity, hypoganglionosis has been rarely reported in literature. The clinical and epidemiological features of HG is similar to HD. When chronic constipation persists despite conservative treatment and exclusion of other differential diagnoses, rectal imaging, manometry and full thickness rectal biopsy should be considered (2). Most adults give a history of abdominal pain, distension and irregular bowel habits. Some however present with severe constipation requiring repeated enemas and complications of the same. Our patient presented with intractable constipation and prior history of sigmoid volvulus. Medical treatment under the presumption of functional constipation is often unsatisfactory. The most frequent diagnosis made on imaging is adult Hirschsprung’s disease when the imaging features are typical. Abdominal radiographs show dilatation of the proximal colon, often to a massive extent with a narrowed distal segment. On barium enema, the proximal segment is massively dilated with fecal residue and a distal narrow transition zone. The transition zone may not be visualised in 20%, due to ultra-short segment disease, especially in children(1). CT is done mainly done to exclude other causes of obstruction and also clearly depicts the transition zone. A much higher transition zone ratio may help in differentiating HD from hypoganglionosis in occasional cases. A transition zone is seen in HD as well as focal type of HG. Surgery is the definitive treatment for HG (7), the principle being removal of the abnormally innervated segments and re-establishment of intestinal continuity. The surgical options for hypoganglionosis can be variable – total colectomy with ileo-rectal anastomosis or localized resection-anastomosis. On the other hand, Hirschsprung’s requires only localised resection in most cases (7).

CONCLUSION

Adult hypoganglionosis is a rare entity with clinical and radiological features mimicking Hirschsprung’s disease. The neuronal innervations disorders also need to be differentiated from other functional conditions. Full thickness rectal biopsy is required to make a definite diagnosis.

REFERENCES
