

Isolated adult hypoganglionosis presenting as sigmoid volvulus: a case report.

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Abstract

INTRODUCTION:

Isolated hypoganglionosis is a rare cause of intestinal innervation defects. It is characterized by sparse and small myenteric ganglia, absent or low acetylcholinesterase activity in the lamina propria and hypertrophy of the muscularis mucosae, principally in the region of the colon and rectum. It accounts for 5% of all intestinal neuronal malformations. To the best of our knowledge, only 92 cases of isolated hypoganglionosis were reported from 1978 to 2009. Isolated hypoganglionosis usually manifests as enterocolitis or poor bowel function, and is diagnosed in infancy or childhood. We report the first case of isolated hypoganglionosis presenting with sigmoid volvulus in a 34-year-old woman.

CASE PRESENTATION:

A 34-year-old Asian woman had progressively increasing abdominal pain and had not passed stool or flatus for two days. A physical examination revealed a distended abdomen with sluggish gut sounds. A computerized tomography (CT) scan demonstrated gross dilatation of the sigmoid colon (maximal diameter 14.3 cm) suggestive of sigmoid volvulus. During emergency laparotomy, sigmoidectomy with a side-to-side colorectal anastomosis was performed. Histopathology of the resected specimen showed occasional ganglion cells and hypertrophied nerve bundles in the muscle layers, suggesting hypoganglionosis. Colonoscopy was performed, and multiple full-thickness biopsies were taken that showed hypoganglionosis of the entire large bowel. Our patient underwent total colectomy with an ileorectal anastomosis. Subsequently our patient reported a dramatic improvement in her bowel function.

CONCLUSIONS:

Isolated hypoganglionosis is a rare cause of intestinal dysganglionosis and cannot be differentiated from Hirschsprung's disease based on clinical presentation. This case report describes an atypical presentation of the disease. A definitive diagnosis requires histopathological analysis of full-thickness intestinal biopsies. Treatment should be tailored to the extent of hypoganglionosis.

Key words; hypoganglionosis, sigmoid volvulus, acetylcholinesterase