CHALLENGES OF HIRSCHSPRUNG'S DISEASE PRESENTING IN AN ADULT: A CASE REPORT

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ABSTRACT

Hirschprung's disease is rare in adults; it is often misdiagnosed or undiagnosed. We present a 34year old man illustrating the clinical characteristic of the pathology, the diagnostic clues and assessing the therapeutic approach. Definitive diagnosis is established on histology specimen from the rectum and the disease was confined to the rectosigmoid .Treatment involved sigmoid colostomy, anterior resection with colorectal anastomosis with a posterior myotomy.

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INTRODUCTION

Hirschsprung's disease is a congenital aganglionosis of the submucosal and myenteric plexuses principally affecting the rectosigmoid or rectal segments to a varying length[1]. It is usually diagnosed in the newborn period affecting 1 in 5000 live births but in rare instances, it is diagnosed in adults. [2]Adult Hirschsprung's disease occurs when the patient is more than 10 years at the time of first diagnosis.

The disease occurs in 2% of the population, the patients have milder disease due to hypertrophied innervated proximal colon compensating for the distal aganglionic rectum[3]. These patients try to relieve constipation by using enemas and cathartics. Constipation or fecal retention will develop when the proximal dilated colon will no longer propel the feces f distally[1-3]. We present a case of a 34 year old man who developed massively distended nonfunctional colon that was treated with sigmoid colostomy, anterior resection with colorectal anastomosis and posterior myotomy.

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CASE PRESENTATION

We present the case of a 34-year old man with chronic constipation dating from childhood and has been managed with dietary modifications. He had emergency sigmoid colostomy for acute intestinal obstruction. Evaluation with barium enema and rectal biopsy revealed megacolon and intestinal aganglionosis which was limited to the rectosigmoid colon. There was past history of achalasia which was treated 10 years prior to presentation by Heller's cardiomyotomy. He subsequently had definitive treatment in form of a low anterior resection and posterior myotomy.

Post operatively he had uneventful recovery and has been opening bowel regularly.

DISCUSSION

The autopsy report published by Frederick Ruysch[4] in 1691entitled''Enormis intestine coli dilatatio'' was the first recorded observation of Hirschsprung's disease. Two centuries later, Hirschsprung[5] reported the disease in 1888. Whitehouse and kernohan[6] in 1948documented the absence of ganglion cells in the myenteric and submucous plexus in all patients with megacolon which forms the pathological basis of the disease. The above finding led to an effective surgical treatment by Swenson and Bill[7] in 1948.

The primary pathological defect is the total absence of ganglion cells of the submucosa l(Meissner) and myenteric (Auerbach's) plexuses in the affected portion of the colon. Human embryos and fetal studies led to the conclusion there was defective migration of ganglion cell precursors of the neural crest into the hindgut[8]. Other theories suggest loss of interstitial cells of cajal which may explain achalazia in some patients with Hirschsprungs disease. The aganglionic segment is contracted while the proximal segment is dilated due to attempts to overcome the obstruction.

The diagnosis of Hirschsprungs disease is based on barium enema[9] which will show a clear cut transition zone between aganglionic distal bowel which is narrow and ganglionic proximal dilated colon. The anorectal manometry typically demonstrates no internal sphincter relaxation in response torectal distension. Rectal biopsy confirms the diagnosis which shows absence ganglion cells, hyperplasia and hypertrophy of nerve fibers, increased levels of the enzyme acetylcholinesterase.

The term adult Hirschsprungs disease is used when the patient is older than 10 years at the time of diagnosis based on different case reviews in the English literature based on total of number cases, clinical presentation, radiological and pathological features. Rosin et al[10] in 1950 documented a case of adult Hirschsprung's in a 54year old physician with a rectosigmoid disease. Miyamoto et al[11] reported a 23 year old man who had a history of chronic constipation that required daily enemas since early infancy. The patient had remained in good health until he experienced severe intestinal obstruction for which an emergency colostomy was performed as seen in our case.

The typical adult patient with Hirschsprung's has a long standing history of constipation since childhood, abdominal discomfort, distension and abdominal pain as seen in our patient[12]. Patient age ranges from 10 to 73 years, and the average age is24.1 years. Half of the patients are younger than 30 years.

Surgical treatment of Hirschsprung's disease consist of resection or exclusion of the aganglionic segment of the colon while avoiding nerve injury and preserving normal anal sphincter function. State[13] in 1952 described the anterior resection of the colon as a therapeutic modality in the management of Hirschsprung's disease in 3 patients aged between 10 to 21. Lynn[14] described myectomy in 1966 for short segment disease which can also be used in combination with laparoscopy for more extensive forms of the disease.

Posterior anorectal myotomy with low anterior resection was the choice in our patient due to the long term results and the procedure has been complication free. However, Doodnath[15]in his review showed that Duhamel[17] procedure was used in 231 patients among 490 patients who have undergone surgery. This technique does not require resection of the aganglionic rectum, but excludes any extensive pelvic dissection that may damage the pelvic sensory nerves. Swenson's procedures

which involves extensive pelvic dissection especially in an adult with increased depth of the pelvis might increase injury to the pelvic nerves leading to impotence, while Soave's endorectal pullthrough[18] eliminates injury to the pelvic nerves but because the depth of the pelvis in adults is more technically difficult with associated major complication when compared with children.

CONCLUSION

This case report highlights our experience in managing this patient, and emphasizes the need for continuing update of information on this disease to health care workers and caregivers in order to improve time to diagnosis.

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