Hirschsprung's disease in an adult

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Abstract Hirschsprung's disease (congenital megacolon) is widely known as a disease of neonatal to childhood. Presentation in adulthood does occur but is rare. To date only 49 cases have been reported in the literature. We presented an additional case of adult Hirschsprung's disease (HD) to enrich the literature and to create an awareness of its existence in adults presenting with chronic constipation from childhood.

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The classic description of Hirschsprung's disease (HD) by Hirschsprung in 1888 was a disease in childhood that caused "massive dilatation and hypertrophy" of the colon. He also remarked that it was not confined to children and believed that some patients may have a milder form and hence survive till adulthood. It was not until the 1950s that the first histologically proven adult case of Hirschsprung's disease was found in a 53-year-old physician. Since then about 49 adult cases between 18-74 years have been described.

Case Report A 33-year old, nulliparous female presented with chronic constipation from childhood. Bowel evacuation was achieved with various laxatives and occasional colonic washouts at the local hospital. She had intermittent lower abdominal pain but no distension and no history of vomiting or diarrhea. No previous investigations had been carried out. Clinical examination revealed no significant findings. Her biochemical profile was also normal. Barium enema showed narrowing of the rectum with a distinct transitional zone leading to a distended sigmoid colon (Fig. 1). The descending colon was only minimally distended. Anorectal manometry demonstrated absence of internal sphincter relaxation and high internal anal pressure (Fig. 2). Endoscopic rectal biopsy confirmed absence of ganglion cells (Fig. 3). Patient underwent surgical exploration where the sigmoid part of the colon was hugely dilated. Preoperative, full-thickness biopsies were taken which confirmed absence of ganglion cells between 2-15 cm from anal verge. Most of the rectum and part of the sigmoid colon were resected. Postoperatively, there were no complications, patient's bowel habit returned to normal and she was doing well in follow up at four months.

Discussion The most constant presenting symptom is long standing constipation. Patients achieve bowel evacuation by increased laxatives and regular enemas. Our patient had this classic presentation. Other less common features including abdominal distension were abdominal pain and vomiting. Constipation is consequent on the pathophysiology of the disease which involves absence of the ganglion cells in the Auerbach's and Meissner's plexus in the affected segment of the bowel. The process is thought to be due to arrest of the usual cranio-caudal migration of primitive neuroblasts, hence, its association in children with other abnormalities of the neural crest cells like neuroblastomas. The involved segment is variable in length ranging from total colonic aganglionosis to a very short segment of the disease. Nevertheless, 85% of the cases are limited to the descending colon and distal segment. There would appear to be three patterns of sex

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distribution depending on the type of HD. The familial type, which usually involves a long segment of the colon, has an equal sex distribution. Sporadic HD largely involves the rectosigmoid colon and has a male preponderance of about 4:1. Adult HD also largely involves the rectosigmoid, but the sex incidence appears controversial. Hung et al. states a female predominance, while Crocker and Messmer in their review had a male to female ratio of 4:1.

Initial evaluation is usually by barium studies where adequate projection is crucial to obtain the transitional zone. The examination is then discontinued to avoid barium impaction. A review of the reported cases shows that 83% would have distal narrowing and proximal dilatation at the rectosigmoid junction. However, a dilated colon without narrowing was seen in 14% of cases and one case had normal looking barium enema with total colonic aganglionosis. Our patient had the typical presentation of rectal narrowing and proximal sigmoid dilatation. Manometric studies are vital to help in differentiating HD from acquired megacolon since the treatment of choice in the former is surgical and the latter medical. The characteristic findings in HD at manometry are

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**Fig. 1** - Barium enema: Rectum is contracted and the sigmoid colon is grossly distended. A distinct transitional zone exists between them.

**Fig. 3** - Submucosal part of a rectal biopsy taken at 4 cm above the anal valve. There is a complete absence of ganglion cells in the submucosal plexus. Note the muscularis mucosa in the upper left of the photograph. A group of nerve fibers is seen in the middle. Hematoxylin and Eosin stain (X400).
Fig. 2 - Recordings of ano-rectal pressure before and after intrarectal balloon inflation (I) with 90 ml of air. Top tracing (channel 3) showing rectal pressure before and after balloon inflation. Channel 4,5 showing internal sphincter pressure. Note that rectal inflation induces no relaxation of internal sphincter with high basal resting pressure of 70 mmHg.
failure of the internal sphincter to relax following rectal balloon distension and high internal sphincter basal pressure.\textsuperscript{12} Histology provides the final confirmation and helps in the direction of extent of surgical excision. However, recent studies\textsuperscript{13} recommend the application of immunohistochemical diagnosis to further enhance the diagnosis. Several methods of surgical treatment are now available, the most acceptable\textsuperscript{14} being anorectal myectomy with low anterior resection, the Duhamel-Martin procedure and the Soave endorectal pull-through procedure. Our patient had the pull-through procedure.

In conclusion, an adult case of HD is presented. We stress the possibility of its existence in an adult with constipation from infancy. The typical findings of rectal narrowing with proximal distension are discussed. Its existence with general colonic distension or normal looking barium enema should also be remembered. The importance of manometric studies and biopsy confirmation are also stressed.

REFERENCES


ملخص:

من المتعارف عليه أن داء هيرشبرنغ يصاب به الرضع والأطفال، مع أن إصابة البالغين نادرة إلا أنه عند الاطلاع على المراجع وجد أن مجموع ما نشر في العالم لم يتجاوز التسعة والأربعين حالة فقط. ولقد قمنا بنشر هذه الحالة لنلفت الانتباه إلى وجود المرض وخصوصاً عند البالغين الذين يعانون من إمساك مزمن منذ الطفولة.

ملفتاح الكلمات: داء هيرشبرنغ.