Case Report

Hirchsprung's disease in a young adult: Report of a case and review of the literature

Mohan Kant Thakur[1*], PritamDas[1], Saugat Das[2], Ksh. Raju Singh[3], ChongthamArun Kumar Singh[4], Laishram Jaleshwar[5]

[1,2]PGT, [3]Assistant Professor, [4]Professor, [5]Senior Registrar, Regional Institute of Medical Sciences, Imphal, Manipur, INDIA.
Email: ttomkant@gmail.com, tomkant_18@yahoo.in

Abstract

Hirschsprung's disease (HD) in adults is rare and often undiagnosed or misdiagnosed. We report a case of HD in a 26-year-old male who had a history of chronic constipation that required laxatives and enemas since early childhood. He developed severe intestinal obstruction and presented to the emergency department with significant abdominal distension. A diversion loop ileostomy was done and full- thickness rectal biopsy taken which showed no ganglion cells. More than two years later he presented with features of obstruction again and desired a definitive surgery. Duhamel's operation was done with revision of the Ileostomy. Histological examination of the resected sigmoid colon and first part of rectum showed reduced ganglion cells in the submucosal and myenteric plexus. His postoperative course was uneventful with complete resolution of the symptoms. Hirschsprung's disease should be considered in adults who have long-standing and refractory constipation

Keywords: Hirschsprung's disease, Constipation, Megacolon, Adult.

[*]**Address for Correspondence:**
Dr. Mohan Kant Thakur, PGT, Regional Institute of Medical Sciences, Imphal, Manipur, INDIA.
Email: tomkant_18@yahoo.in
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INTRODUCTION

Hirschsprung's disease (HD) is a congenital illness in which there is a lack of intrinsic nerves (ganglion cells) in the distal segments of the intestinal tract[1]. These abnormal segments produce mechanical obstruction because of failure to relax during peristalsis[2]. Hirschsprung's disease always starts in the anal verge, but the length of the segment without ganglion cells (aganglionic) varies: it is limited to the rectum and sigmoid in 75% of patients; involves the whole colon in 8%; and rarely involves the small bowel.

HD was first described by Harald Hirchsprung, a Danish physician, in 1886[3]. It was a description of few cases of constipation in children in association with dilatation of Sigmoid colon. The dilated sigmoid colon was thought to be the offending factor then. It was later realized that the offending factor was actually a functional obstruction in the rectum due to the absence of ganglion cells. Dilation of the sigmoid was an after effect of a distal obstruction.

Usually, HD patients are diagnosed early in life and require surgical treatment. Hence adult HD is an uncommon diagnosis. Some patients may go undiagnosed till adulthood because the colonic region proximal to the distally obstructed segment assumes a compensatory role by dilation. These cases in which diagnosis is established in adulthood, specifically after ten years of age, are known as "Adult HD"[4,5]. Herein, we report a case of adult HD treated by modified Martin-Duhamel procedure, a feasible surgical option for adult HD.

CASE REPORT

A 26 years male presented in the OPD with the presenting complaints of abdominal distension and with a diversion ileostomy per abdomen.More than two years back he presented with similar complaints of distension and constipation. X-ray was suggestive of dilated bowel loops. Laparoscopy was done and dilated bowel loops

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were confirmed. He underwent an emergency laparotomy, operative findings were consistent with radiography so a diversion loop ileostomy was created to relieve obstruction and due to high suspicion of a motility disorder full thickness rectal biopsy was taken. The then operating surgeon formed the ileostomy in the left iliac fossa. The histopathological examination of the biopsy specimen showed absence of ganglion cells. Patient was discharged then by the treating doctor. Post-operative barium enema was suggestive of grossly dilated sigmoid colon and narrowing of recto-sigmoid region of 3 cm in length. A diagnosis of Hirchsprung's disease was made.

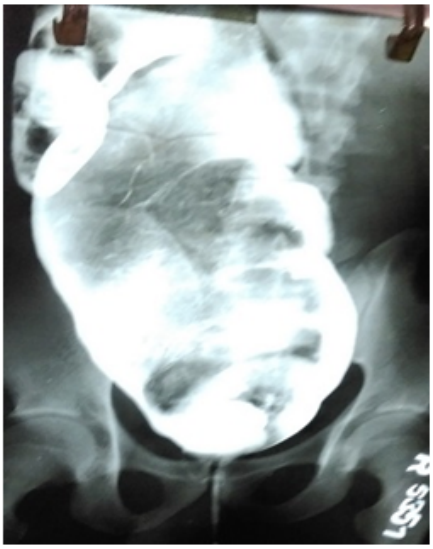

Figure 1 Barium enema- dilated sigmoid colon

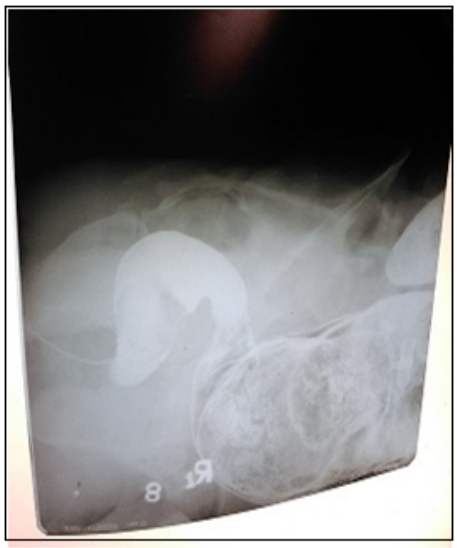

Figure 2: Barium enema

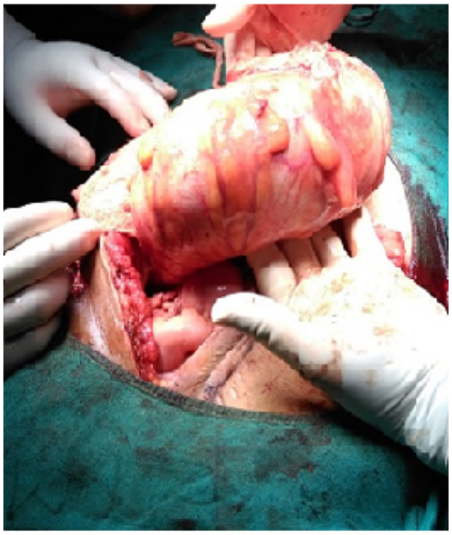

Figure 3: Dilated colon

After about two and a half years, he presented with the similar features in our OPD. On abdominal examination, abdomen was found distended without any signs of peritonitis. Bowel sounds were exaggerated. Digital rectal examination was unremarkable. Rest of the systemic examination was not suggestive of any significant finding. Biochemical profile was within normal range. Abdominal X-ray showed dilated small bowel and fecal loading of large bowel. Definitive surgery was planned as was the choice of the patient. Midline laparotomy was done and massively dilated sigmoid colon was observed with dense adhesions to anterior and lateral abdominal walls. Duhamel's operation was done and the ileostomy site was revised to the right iliac fossa. There were not any significant post-operative events and the patient was discharged on the 10th post-operative day. Patient had been followed regularly since then on monthly intervals.

DISCUSSION

Neural crest cells migrate cranio-caudally to innervate the entire gut (enteric nervous system). Failure of this migration in the intra-embryonic life results in aganglionosis, which may be classical or recto-sigmoid(short-segment) , long segment HD or total colonic aganglionosis (Zuelzer-Wilson syndrome). The disease usually presents in infancy and rarely in adulthood.The primary defect in adult Hirschsprung's disease is identical to that seen in infancy[6,7]. Adults usually present with a long history of constipation and recurrent episodes of abdominal distension and discomfort. Contrast enema and rectal biopsy are the two most important investigations[8].

A typical transition zone is seen on barium or iopaque enema in HD, with a small calibre aganglionic rectum, a funnel shaped hypo-ganglionic area (the transition zone) and a dilated proximal colon which is normally ganglionated. Barium enema is not as sensitive or reliable as rectal suction biopsy in ruling out Hirschsprung's disease[9]. Rectal biopsy is diagnostic as it may show absence of all the plexuses in the mucosa, submucosa and both the muscular plexuses, high acetylcholine-esterase activity and monoclonal antibodies against certain neural components, e.g., D7[10].

Chief management is surgery and for reconstruction of short segment HD, pull-through procedures are more popular. This includes Duhamel, Swenson and Soave procedures[11,12,13]. Of these, Duhamel's procedure is usually considered the best procedure as it retains the rectum and hence the rectal sensation, storage and better bowel function, thereby reducing the post-operative morbidity and producing better results in adults[14-18].

Major complications are usually not encountered in adult patients, although anastomotic leak, stricture, fistula, pelvic abscess and necrosis or retraction of colon may

occur[19]. Post-operative counselling for increased fibre content in diet is important as constipation and bowel stasis increase the risk of enterocolitis[20,21].

CONCLUSION

Hirchsprung's disease in adults is a rare incidence and hence its diagnosis is challenging and requires high index of suspicion as it can easily be misdiagnosed due to overlapping symptoms. Correct diagnosis and adequate management definitely improves the quality of life of the patient.

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