



Hypertrophic Megacolon in a 1-year-old child with Hirschsprung's Disease: Case Report and Literature Review



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Abstract

Hirschsprung's disease (HSCR) is a common cause of paediatric intestinal obstruction. It is a functional colonic obstruction due to lack of ganglion cells in the affected segment of colon. Symptoms range from intestinal obstruction to chronic progressive constipation in older children. A 1-year-old infant was brought to Khartoum Teaching Hospital with abdominal distension, difficulty passing stool and chronic constipation treated by regular enema without improvement. On examination, patient looked ill and emaciated. Diagnosis of HSCR disease was established and patient was planned for operative management. The clinical presentation and management of chronic HSCR were briefly discussed in this report.

Keywords: Hirschsprung's disease; Hypertrophic megacolon; 1-year old, Child

Introduction

Hirschsprung's disease (HSCR) is caused by the failure of ganglion cells to migrate from the colon during gestation. The different lengths of the distal colon cannot relax, resulting in functional colonic obstruction. The disease usually appears in early infancy, although some patients experience severe and persistent constipation later in life. Symptoms include difficult defecation, poor feeding, poor weight gain, and bloating. This disease can be familial or spontaneous, and multiple factors appear to play a causative role in HSCR disease [1].

Patients diagnosed later in life with chronic symptoms often have short-segment variant of the disease, and their clinical manifestations often include chronic constipation, bloating, vomiting and growth failure [2]. Older children who suffer from long term obstructive symptoms, are resistant to conventional treatment options and may require daily enema treatment.

Enterocolitis and ruptured colon are the most serious complications associated with the disease and the most common causes of mortality related to it [3]. Enterocolitis occurs in 17% to 50% of infants with HSCR disease, and the most common causes are intestinal obstruction and residual ganglion enteritis. For many years after corrective surgery, babies must continue

to be closely monitored for enterocolitis, as infections have been reported up to 10 years later [1].

Case Report

A 1-year-old infant was brought to Khartoum Teaching Hospital with abdominal distension, difficulty passing stool and chronic constipation treated by regular enema without improvement. His condition started since birth when patient delivered at home by non-vaginal delivery, of full term pregnancy which passed uneventful, he cried immediately and started to breastfeed. Baby failed to pass meconium during first 48hrs after birth.

On examination; patient looked ill and emaciated. Digital rectal examination showed hard stool with gush of fluid upon finger removal. Plane abdominal X-ray showed dilated colon with fecal impaction. Contrast enema study showed huge dilated rectosigmoid with transitional zone and collapsed distal rectum. Interestingly, enema study also showed a thick hypertrophied colon with very big old stool that appeared to be hardened.

Patient was admitted, cannulated and given intravenous antibiotics, then rectal wash by saline 10-15ml/kg for one week was done. Thereafter, he was prepared for colostomy and

biopsy. Histopathological report showed typical features of distal aganglionic bowel segment.

Patient was operated under general anesthesia and aseptic condition, left iliac fossa grid iron incision was made, and bowel explored. Three seromuscular biopsies was taken from the collapsed part of rectosigmoid colon, stoma site and the dilated

part, then loop colostomy was created. Patient recovered well from anesthesia. Three day post-operative stoma were adequately functioning and patient was discharged home and planned for Swenson pull through operation after 6 months. The operation was done successfully and follow up went uneventfully (Figures 1-3).



Figure 1: Semitendinosus and gracilis tendon harvesting.



Figure 2: Radiographic abdominal lateral view of water-soluble contrast enema.



Figure 3: Loop colostomy for bowel defunctioning.

Discussion

Aganglionic disease is the main etiology of HSCR. The full migration, proliferation, differentiation, survival and apoptosis of neural c cells all contribute to functional ENS. Any interference in these processes will cause HSCR disease phenotype. In 80-85% of HSCR cases, the aganglionic area is limited to the rectum and sigmoid colon (short-segment disease). Long-segment disease can occur in up to 20% of cases and has the characteristics of aganglionic disease that extends to the proximal end of the sigmoid colon. Total colonic aganglionic disease is rare, occurring in 3-8% of HSCR patients [3].

About 80% of cases present in the first months of life, with dysmotility, poor diet and progressive abdominal distention. Up to 90% of babies with HSCR disease do not pass meconium within the first 24 hours after birth [1]. Older children present with constipation that doesn't respond to regular treatment, these children usually don't develop normally and are often thin and malnourished.

A retrospective study was conducted on 64 patients of HSCR in Sudan, with male: female ratio 5:1, average age at first presentation was nine days and most patient were from central Sudan. Patients present commonly with delayed passage of meconium and constipation, moreover, emergency presentation was encountered in 21.9% of cases. Complications were reported most frequently on the 13th day following surgery with colostomy prolapse. The age of patients at Pull Through Procedure was ranging from seven to seventy two months and the mean body weight was around 12 kg, these factors were found to play an important role on surgical outcome and length of hospital stay [4].

The diagnosis of HSCR can be done in a variety of ways. However, the preferred diagnostic procedure of choice is a contrast enema. This will define the transition zone between the normal (dilated) bowel and the narrow aganglionic bowel. This transition zone can be seen in 70% to 90% of cases. Routine x-rays show dilation of the intestine, and anorectal manometry can also help the diagnosis [3]. In rectosigmoid HSCR the location of radiographic transitional zone correlate accurately with the level of aganglionosis in majority of cases, but fail to be accurate in long-segment or total colonic disease [5]. The gold standard for diagnosing HSCR is rectal biopsy. Submucosal rectal aspiration biopsy can be performed without anesthesia. A biopsy sample is analyzed to look for the loss of ganglion cells and hypertrophic nerve trunks. Biopsy samples can be stained to increase the activity of acetylcholinesterase, thereby aid in the diagnosis. If aspiration biopsy does not provide an accurate diagnosis, a full-thickness biopsy should be performed [3]. Simple modified technique can improve the adequacy of rectal suction biopsy, and wall suction with Noblett forceps is more likely to produce an adequate specimen and decrease the need for repeated biopsy [6].

Until now, the only treatment for HSCR is surgery. Due to malnutrition or sepsis after bowel perforation, lack of surgical treatment for HSCR can be fatal. Although surgery is the conventional treatment for HSCR patients, the results of surgery vary widely and vary widely [3]. Traditional treatment by the three stage Pull Through Procedure carry disadvantages, such as stoma problems and might triple hospital stay time. Thus, one stage operation is more preferred option for neonates [7].

Surgical treatment aims to remove the ganglion intestine and anastomose the normal intestine to the anus, to maintain

the function of the sphincter. The main technologies include total Transanorectal removal (TERPT) and laparoscopic assisted removal procedures. The modified TERPT procedure has generally better long-term outcomes than the classical approaches [8].

Conclusion

Early diagnosis and treatment of HSCR are essential to achieve better results. Initial diagnosis can be made by water-soluble contrast enema, but the gold standard for HSCR is rectal biopsy, and the mainstay of treatment is surgery. Chronic form of disease usually present with chronic constipation that doesn't respond to regular enema, imaging may demonstrate hypertrophied colon with hard stool impaction. In emergencies, the age and weight at the time of removal surgery and the closing time of the colostomy can significantly affect the prognosis and extend the length of hospital stay.

Consent

Written consent was obtained from the patient and his parents to write and publish his case as a report with accompanying radiological images.

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Conflicts of Interest

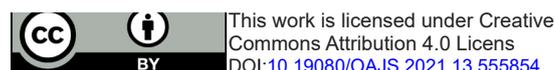
Authors declare that there are no conflicts of interest regarding the publication of this paper.

Authors' Contributions

Authors contributed equally and significantly to the concept and design, drafting, critical revision and final approval of the report.

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