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Case Report

A case report of a pediatric patient with Hirschsprung's disease

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ABSTRACT

The objective is to describe an uncommon case of Hirschsprung's disease (HD), also called congenital megacolon. The lack of neuronal ganglion cells from the submucosal and muscular nerve plexuses throughout a segment of varied length characterizes HD. Approx 94% of HD cases are diagnosed below the age of 5 years. A 3 years old male patient was admitted to the male surgery ward under a pediatric surgeon with the chief complaints of chronic constipation since birth with a history of delayed passage of meconium. The patient was diagnosed with a case of HD at the age of 1 year for which he was operated on colostomy was performed with multiple biopsies from narrowed rectosigmoid, from the transitional zone, and from colostomy stoma which confirmed the decision to make colostomy at the ganglionated segment of the colon based on the clinical picture during surgery. Biopsy report of narrowed rectosigmoid segment showed the absence of ganglion cells, while colostomy stoma and dilated proximal colon showed the presence of ganglion cells. The patient underwent a surgical procedure named pull-through surgery for HD, a martin's modification of the Duhamel operation.

Keywords: Congenital megacolon, Chronic constipation, Rectosigmoid, Colostomy

INTRODUCTION

Hirschsprung's disease (HD), often called congenital megacolon, was innovated by Harald Hirschsprung, a Danish doctor, he made the first description of it in 1886.[1,2] The lack of neuronal ganglion cells from the submucosal and muscular nerve plexuses throughout a segment of varied length characterizes HD.[3-6] The incidence of the disease is 1 in every 5000 live births.[7] In the adult population, 5% of cases are identified.[2] It happens as a result of a lack of or diminished number of intramural ganglion cells particularly in the distal colon.[8] This disease is having a 3:1 total male: female ratio.[7,9] Though diagnosis of HD in adults is rare, the disease commonly affects children.[4,5] HD is treated surgically, which involves the removal of the portion of the colon that lacks nerve cells. The disease distally starts from just above the dentate line and extends proximally involving the rectum, rectosigmoid, and sigmoid colon in many cases. Sometimes the aganglionosis may extend cephalically to get involve descending colon, splenic flexor, transverse colon, hepatic flexor, ascending colon, or even the caecum (Total colonic aganglionosis). Rarely the disease may extend and involves the terminal ileum, entire ileum, or even the jejunum, but the stomach and duodenum are never involved in the aganglionosis.

It varies in many children; the disease presents with chronic constipation. In a few patients, the disease manifests in neonates as neonatal intestinal obstruction.[10] Many patients have recurrent abdominal distension, which is relieved by rectal examination or enemas. A full-term neonate, who does not pass meconium within 24-48 hours, or a premature who has not passed meconium in the first 72 hours, should be suspected of having HD until proven otherwise. The diagnosis is

confirmed by a lower GI contrast study. Rectal biopsy or rectal manometry, where rectal biopsy is confirmatory.[11]

Diagnosis

Based on a combination of data HD is diagnosed by the following techniques[12] 1. Anorectal manometry (ARM), 2. Radiographic studies-plain abdominopelvic X-rays, contrast enema (CE). 3. Biopsy-rectal suction biopsy (RSB), full thickness, or submucosal rectal biopsy. 4. Genetic studies-The disease also shows a genetic association.

Treatment

HD is treated surgically, which involves the removal of the portion of the colon that lacks nerve cells, but before definitive surgery, preliminary diversion in form of colostomy is usually needed with multiple extra mucosal colonic biopsies.[13]

CASE REPORT

A 3-year-old male patient was admitted to the male surgery ward with a colostomy in the left flank as a case of HD for further definitive surgery (Figure 1). History was not contributory. No similar complaints were seen in siblings. Renal and liver biochemical reports were normal. A biopsy report of previous surgery showed absent ganglion cells in narrowed rectosigmoid segment. While colostomy stoma and dilated proximal colon showed the presence of ganglion cells in the submucosal and myenteric plexus. Based on history examination and investigation final diagnosis of HDs was made. Under the guidance of a pediatric surgeon, the person was treated. The patient underwent a surgical procedure by technique of martin's modification of Duhamel operation (Figure 2).

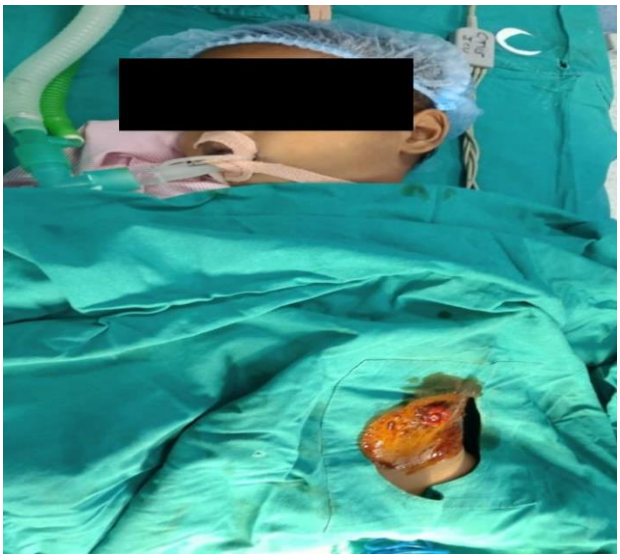

Figure 1: Colostomy for bowel dysfunction.

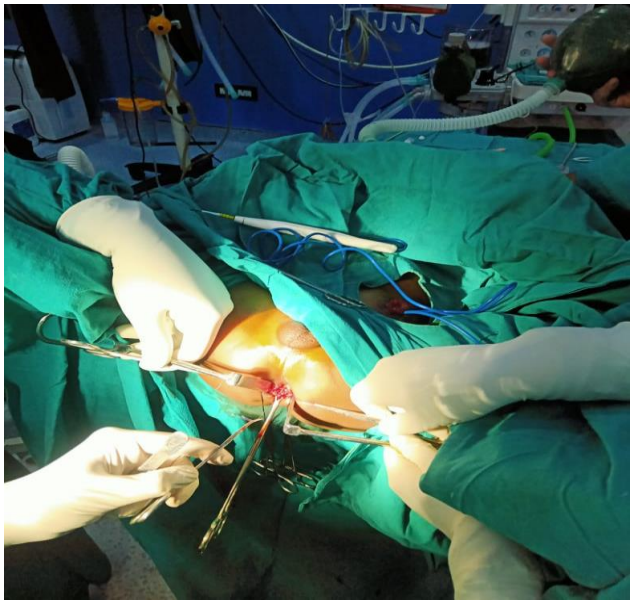

Figure 2: Pull through surgery for HD.

DISCUSSION

According to the WHO, HD affects about 1 in every 4000-5000 live births. However, the incidence rate in adults is unknown or late age. Ratio of male-to-female is 3:1.[7] In adulthood, the incidence of the disease is more in females in a ratio of 1:3.[14] The median age is 4.85 years.[15] In about 80% of cases, constipation, a poor diet, and progressive abdominal distention are the main presenting feature of the disease. X-ray abdomen in standing posture, and low gastrointestinal contrast studies, are used in the diagnosis of the disease but the accuracy is less than 75%. For confirmation, a Rectal biopsy should be performed.[16] In the cases of, those who are presenting with signs of acute intestinal obstruction and undergoing exploratory laparotomy should have multiple biopsies and colostomy or ileostomy on the innervated segment of the bowel based on clinical findings during surgery. If facilities are available, frozen section biopsies may be considered for choosing the right segment of the bowel for exteriorization. In other cases, a rectal biopsy is the deciding diagnostic tool for further steps of the surgical procedure. The aim to cure the disease is to remove the aganglionic segment of the bowel and get pull the innervated bowel for a new rectum. This can be achieved by complete removal of the aganglionic bowel or by leaving some part of the aganglionic rectum to protect pelvic neural structures by vigorous pelvic dissection and getting anastomose of the pulled ganglionic bowel to the residual native aganglionic rectum. A third technique may be used to leave the aganglionic rectum as a seromuscular sleeve without mucous and get pull the ganglionated bowel from within this sleeve. The patient was 3 years old male admitted to the male surgery ward with the chief complaint of chronic constipation since birth with a history of delayed passage of meconium. The patient was diagnosed with a case of HD at the age of 1 year for which he was operated on, colostomy was

performed with multiple biopsies from narrowed rectosigmoid, from the transitional zone, and from colostomy stoma which confirmed the decision of making colostomy at the ganglionated segment of colon. The patient received care from a team of health professionals including clinical pharmacists, nursing staff, and pediatric surgeons. After getting proper care from the health care professionals his symptoms got relieved and he must take medicines as prescribed. His parents were counseled appropriately to take care of his child and follow the instructions of health care professionals.

CONCLUSION

In this report, we discussed an uncommon case of HD. Our study reports that the symptoms of the patient get relieved after taking proper medication and the surgical procedure done by a pediatric surgeon. This case highlights the importance of rectal Biopsy in detecting the condition and it must be noted for improving outcomes of surgical treatment. Early surgery is indicated to relieve the symptoms and to cure the child.

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