Case Report

Hirschsprung disease in an adult: Too late to diagnose?

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ABSTRACT

Adult Hirschsprung disease is a rare disorder of the hindgut that is frequently misdiagnosed as refractory constipation. About 94% of Hirschsprung disease cases are diagnosed before the patient reaches 5 years of age, however, on a rare occasion, mild cases may go undiagnosed until he or she reaches adulthood. We are presenting the case of an 18-year-old male who came with acute intestinal obstruction with a history of refractory constipation since childhood. Exploratory laparotomy with sigmoid diversion colostomy was done along with full-thickness rectal biopsy. Rectal biopsy showed the absence of ganglion cells. The patient got symptomatically relieved. Therefore, a high suspicion of Hirschsprung disease should be kept in adults having a history of repeated episodes of obstruction since childhood.

Key words: Adult Hirschsprung disease, Aganglionosis, Recurrent constipation

Hirschsprung disease, also known as aganglionic megacolon [1], was first reported by the Danish physician, Harald Hirschsprung in 1886 [2]. Its incidence is 1 in 5000 live births [3]. Approximately 50–60% of the cases are diagnosed in the 1st month of life [4]. Adult-onset Hirschsprung disease is diagnosed after 10 years of age [5]. Approximately 5% of cases are diagnosed in the adult population [2]. It occurs because of an absence or reduced number of intramural ganglion cells in a segment of distal bowel [6]. An adult usually has a history of refractory constipation which he generally treats himself with cathartic agents. The diagnosis is missed because of low suspicion. It is confirmed by rectal biopsy showing the absence of ganglion cells on histopathological examination (HPE). Management is surgical depending on the age of presentation and segment of bowel involved [4]. The following case has been reported because of the rarity of such a disease and the difficulty of reaching the correct diagnosis, thereby providing the appropriate definitive management.

CASE REPORT

An 18-year-old male presented to the surgery emergency with complaints of pain whole abdomen and non-passage of flatus and feces for 2 days. The pain abdomen was acute in onset, severe in intensity, and colicky in nature. There was no radiation of pain to the back and no exaggerating, or relieving factors. This was associated with multiple episodes of low-volume vomiting throughout the day which was bilious but non-projectile and not blood tinged. There was no relief of pain with vomiting. The patient had several previous episodes of intestinal obstruction since childhood which got relieved with laxatives. There was no history of fever, night sweats, blood in stool, loss of weight, or appetite. There was no previous history of surgery or tuberculosis in the past and no history of tuberculosis in his family. There was no delay in developmental milestones.

On examination, the patient was conscious and oriented to time, place, and person. He was dehydrated. His blood pressure was 92/64 mm of Hg and pulse rate was 112/min. There were no pallor, pedal edema, or cervical lymphadenopathy. Abdominal examination revealed distension with the fullness of flanks and visible peristalsis. There was a generalized tenderness. His hernial sites showed no cough impulse. There was no fluid thrill and the whole abdomen was tympanic on resonance. His bowel sounds were exaggerated. The digital rectal examination revealed an empty rectum with no fecal staining and no mass. The erect X-ray abdomen showed multiple air-fluid levels. The large bowel loops were distended but the rectum had no air (Fig. 1).

History and examination were more suggestive of a large bowel obstruction which was confirmed by the level of obstruction on X-ray. This is a case of acute large bowel obstruction in a young male with repeated episodes of obstruction in the past. Therefore, the differential diagnosis of sigmoid volvulus and Hirschsprung disease was kept.

After adequate resuscitation, the patient underwent exploratory laparotomy. Intraoperatively, he was found to have massively

Access this article online	
Received - 06 October 2020 Initial Review - 21 October 2020 Accepted - 24 October 2020	Quick Response code
DOI: 10.32677/IJCR.2020.v06.i11.003	

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dilated large bowel loops up to the upper rectum (Fig. 2). A full-thickness rectal biopsy was taken and a sigmoid diversion colostomy was done to relieve the obstruction. HPE showed an absence of ganglion cells in the rectal wall (Fig. 3).

Following surgery, the patient was immediately relieved of obstruction. His stoma became functional on the evening of the surgery. He was allowed oral intake on postoperative day (POD) 1. He was discharged on POD3 with a functional and healthy stoma. He followed up a week later for suture removal. He had no complaints and his stoma was healthy and functional. The patient was worked up for definitive management but subsequently got lost to follow-up.

DISCUSSION

The incidence of Hirschsprung disease is 1 in 5000 live births [3]. The incidence rate is not known in adults because it is overlooked at such a late age [6]. It is more common in males with a male-to-female ratio of 3:1 [3]. In adult-onset Hirschsprung disease, females are more commonly affected with a male-to-female ratio of 1:3 [4]. The median age at diagnosis is 4.85 years [7]. A review of literature by Miyamoto *et al*. found that the mean age of adult-onset disease was 24.1 years with a range of 10–73 years [6].

This disorder is characterized by the absence of intramural ganglion cells in the submucosal (Meissner) and myenteric (Auerbach) plexus in the affected segment of the bowel. There is a defective craniocaudal migration of neuroblasts originating from the neural crests during the first 12 weeks of gestation, resulting in functional intestinal obstruction. Adult onset is likely because the colonic region proximal to the distally obstructed segment assumes a compensatory role. At some point, the dilated proximal colonic segment may decompensate secondary to distal obstruction and patients experience rapidly worsening constipation [5].

Most cases are sporadic. 10% of cases are familial. Half of the familial cases and 15% sporadic cases are associated with gene mutations which inactivate the RET (ReArranged during Transfection gene) receptor for tyrosine kinase on chromosome 10q [4]. About 10% of cases are associated with Down's syndrome. Hirschsprung disease is autosomal dominant with incomplete penetrance. Therefore, the modification of genes and environmental factors is important as well. This condition is also linked with MENIIA, congenital deafness, hydrocephalus, bladder diverticulum, Meckel's diverticulum, imperforate anus, ventricular septal defect, renal agenesis, cryptorchidism, Waardenburg syndrome, neuroblastoma, and Ondine's curse [8].

The lower limit of aganglionosis or hypoganglionosis is the internal anal sphincter which is constant [9]. The upper limit is variable. On this basis, Hirschsprung's disease is classified into short-segment disease when the aganglionic segment does not exceed the sigmoid colon and long-segment disease when it outruns the sigmoid colon. About 80% of cases are short-segment disease. It is called ultrashort when it involves the distal part of the rectum, in 2–3% of cases [4].

Patients present with long-standing recurrent constipation, abdominal distension, and pain. They may have a history of fecal incontinence when the liquid stools bypass the distal fecal impaction. The patient may have associated growth and mental retardation [5]. These cases mostly have a history of regular use of cathartics to relieve symptoms. On examination, patients have a distended abdomen with visible peristalsis and possibly palpable fecal masses.

Abdominal radiograph shows a massively distended proximal colon and a small narrow distal segment. X-ray is positive in 80–85% of cases [9]. When the diameter of the cecum is more than 12 cm, ascending colon more than 8 cm or rectosigmoid

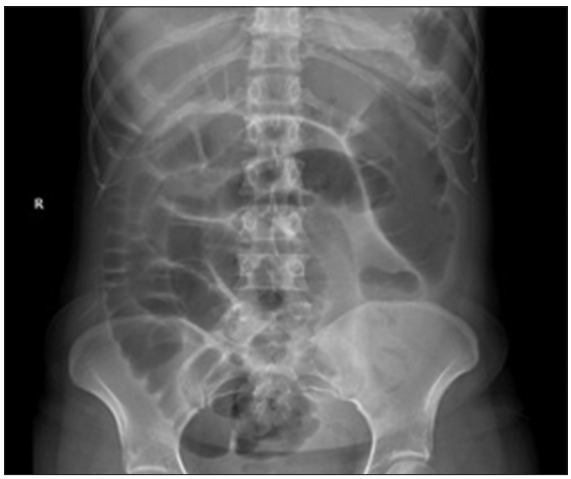

Figure 1: X-ray abdomen showing dilated large bowel loops and absent rectal gas shadow

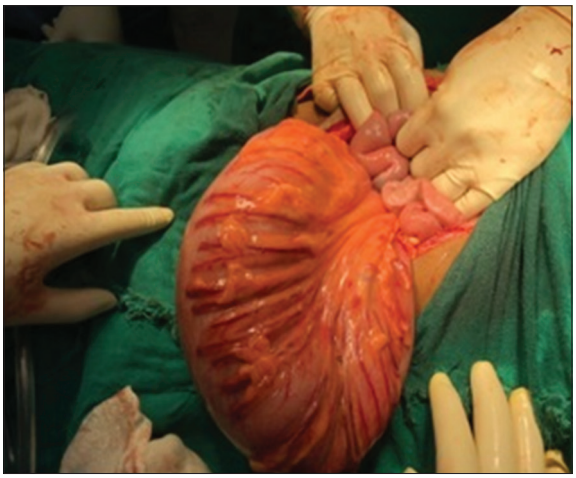

Figure 2: Intraoperative photo showing megasigmoid colon

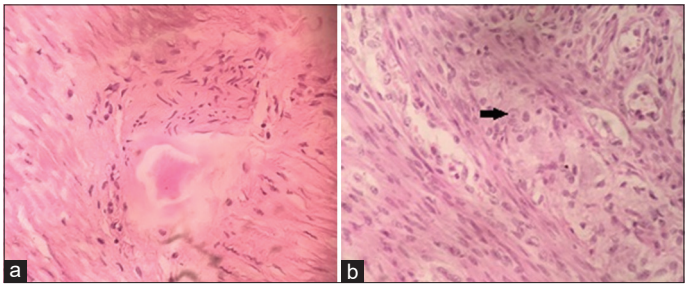

Figure 3: Histopathological examination showing (a) absence of ganglion cells; (b) normal rectal wall (arrow showing ganglion cell)

more than 6.5 cm, megacolon should be considered [7]. A barium enema may show the classical three zones – proximal dilated bowel, affected transitional zone, and distal contracted bowel [5]. It may show an irregular saw-tooth appearance with a cone-shaped transition zone and reversal of rectosigmoid diameter ratio [10]. Barium enema has a sensitivity and specificity of 70% and 83%, respectively [9]. Contrast-enhanced computed tomography views the transition zone and excludes other diseases like colorectal carcinoma [1]. Hirschsprung disease is ruled out by the presence of rectoanal inhibitory reflex on anorectal manometry [4]. This has 91% sensitivity and 94% specificity. The marker of this disease is antibodies against Calretinin. This test has 100% sensitivity and specificity. The gold standard for diagnosis is full-thickness rectal biopsy with 100% sensitivity and specificity [9]. The rectal suction biopsy should be taken 1.5 cm above the dentate line because the distal rectum normally does not have ganglion cells. Full-thickness biopsy is indicated if no hypertrophic nerve trunks are found [8]. HPE may show an increase in non-myelinated nerve fibers in the submucosa and muscle layers [4].

The differential diagnosis of megacolon should include Hirschsprung's disease, intestinal neuronal dysplasia, acquired megacolon, Chagas disease, chronic intestinal pseudo-obstruction, and toxic megacolon due to infection or inflammation [9].

Before surgery, serial rectal irrigations help to decompress the bowel and prevent enterocolitis. The surgical procedure for children can be applied to adults with some modifications. Several approaches exist for treatment. Swenson procedure removes the rectum and does a coloanal anastomosis. Duhamel is a retrorectal transanal pull-through procedure while Soave's technique does an endorectal transanal pull-through. Other procedures include posterior anorectal myectomy and low anterior resection [11]. One-stage approach is feasible for adults due to a relatively healthier nutritional status and the ability to use adult gastrointestinal anastomosis stapler [3]. Exploratory laparotomy is required in an emergency in some cases due to acute intestinal obstruction and atypical clinical features. Serial dilatation of anastomosis is required to prevent stricture formation which can be done at home. Postoperatively, a high fiber diet should be followed to prevent constipation and bowel stasis which increases the risk of enterocolitis [8]. Post-operative complications include enterocolitis occurring in 17–50% of cases, constipation in 10% of cases, and fecal incontinence in 1% of cases [8].

CONCLUSION

High suspicion of Hirschsprung disease should be kept in adults having a history of repeated episodes of obstruction since childhood. Such patients present to the surgical emergency and because of misdiagnosis, they fail to be successfully treated.

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Funding: None; Conflicts of Interest: None Stated.

How to cite this article: Panda V, Ansari FM. Hirschsprung disease in an adult: Too late to diagnose?. Indian J Case Reports. 2020;6(11):615-617.