

ADULT PRESENTATION OF HIRSCHSPRUNG'S DISEASE

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ABSTRACT

Young patients presenting with chronic constipation and failure to thrive, should be promptly investigated and looked upon for late presentation of congenital diseases like Hirschsprung disease (HD). This case is reported to highlight rare case of adult presentation of HD who was brought to emergency department with complaints of lower pain in abdomen, flatulence and lethargy. Considering the findings of CT scan suspicion of Adult presentation of HD was given and lateral biopsy confirmed the diagnosis.

Key words: Chronic constipation, failure to thrive.

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INTRODUCTION

Hirschsprung's disease is congenital disorder in which there is complete absence of nerve plexuses located between longitudinal and circular muscles of the affected segment of bowel. The most common site involved is anus, however it can extend proximally to involve variable degree of colon. Patients having Hirschsprung's disease is usually diagnosed in infancy. Their most frequent presentation is failure to pass meconium in first 24 – 48 hours after birth. It seldom happens that, it goes undetected and later presents in adulthood as chronic constipation. Although the pathophysiology remains the same irrespective of the age at which it presents, may it be adult, infant or child. But the reason for the late presentation lies in the fact that the normal bowel loop that lies proximal to the aganglionic segment gets hypertrophied and hence compensates for the obstruction. Until late in the disease when even cathartic and enemas fail to work and leads to worsening of the symptoms and hence initial presentation.¹

CASE PRESENTATION

A 17 years old male was referred to radiology department through emergency department with

history of failure to thrive and chronic constipation associated with increased flatulence, pain in lower abdomen and anemia since early childhood.

Contrast enhanced CT scan of abdomen and pelvis was performed which showed significantly dilated air and fecal loaded large bowel loops from caecum up to the rectosigmoid junction with abrupt transition of caliber seen at rectosigmoid junction. After which the rectum and anal canal was found be collapsed. Based on CT findings a radiological diagnosis of Hirschsprung's disease was made. The patient was urgently referred to specialist with expert opinion, so as to avoid any further delay and to prevent complications.

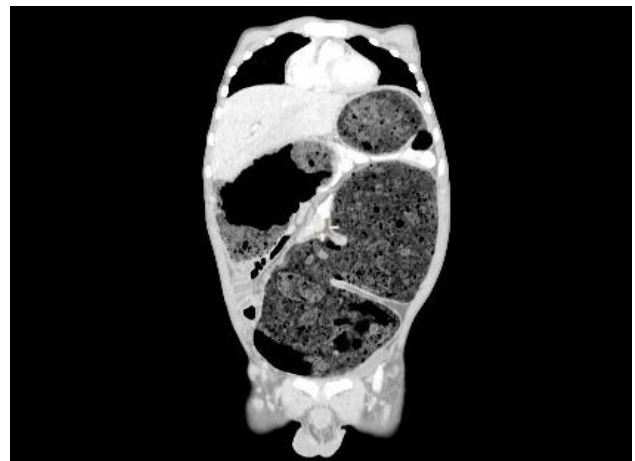


Figure 1: Contrast-enhanced computed tomography of chest, abdomen and pelvis; coronal MPR image, showing an extremely dilated sigmoid colon.

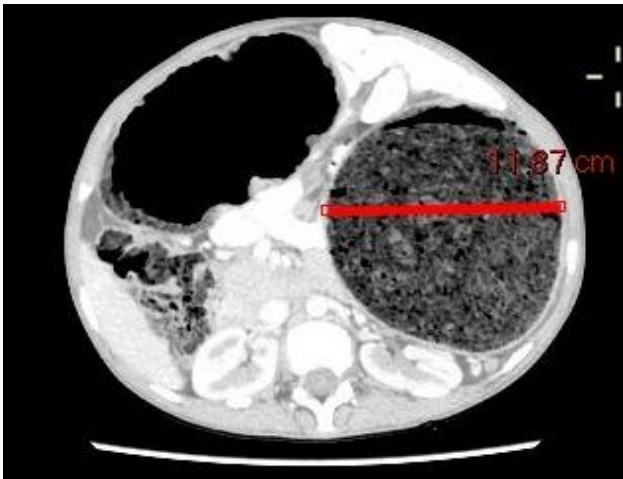


Figure 2: Contrast-enhanced computed tomography of chest, abdomen and pelvis; an axial section at the level of lower poles of kidneys, showing an extremely dilated sigmoid colon having maximum diameter of 11.8 cm in transverse dimension.

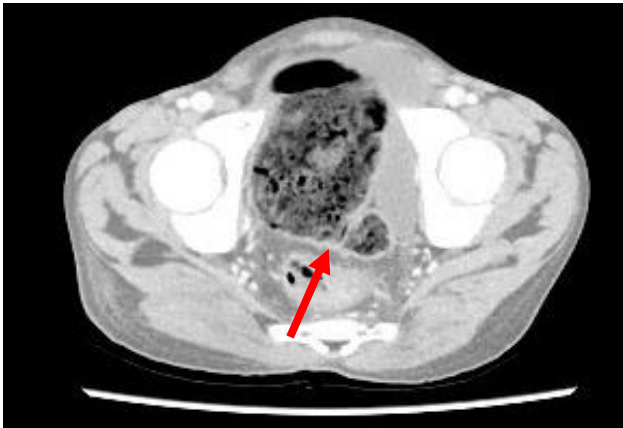


Figure 3: Contrast-enhanced computed tomography of chest, abdomen and pelvis; axial and sagittal images, showing abrupt transition of caliber (red arrows) at rectosigmoid junction.

The patient then underwent exploratory laparotomy and frozen section biopsy was performed, which showed aganglionosis in the collapsed segment. Hence the diagnosis of Hirschsprung's disease confirmed.

The patient then underwent surgery comprising of proctocolectomy and ileoanal pouch anastomosis. Patient was able to tolerate procedure well and showed no immediate postsurgical complications.



Figure 4: Resected segment of bowel loop, showing dilated bowel loops proximally with ischemic necrotic segment distally and a segment of ischemic bowel between the two.

Since adult Hirschsprung disease is a rare cause of chronic constipation; it can easily be missed and therefore it is pertinent to always be keep in it mind in cases of refractory constipation.

DISCUSSION

The Hirschsprung's disease has an overall incidence of 1 in 5000 live births, being more common in male than females 4:1² Failure to pass meconium is the usual presentation in neonatal period with the late presentation of Hirschsprung's disease being chronic constipation or sometimes with a complication like bowel obstruction

The main pathophysiology is complete absence of ganglion cells of the submucosal (Meissner's) and Myenteric (Auerbach's) plexuses in the affected segment of the large bowel. Studies have shown that there is defective migration of ganglion cell precursors of the neural crest into the hindgut.² Some recent molecular studies have associated Hirschsprung's disease to defects in neural crest stem cell function.³

It is commonly associated with Down syndrome in approximately 10% of cases, with neurocristopathy syndromes like MEN IIa, non-neurocristopathy syndromes and with neuroblastomas.^{4,5}

It is anatomically divided into 4 main types. The short segment type being the most common type involves the rectum and distal sigmoid colon, followed by the long segment type involving extending up to the splenic flexure or transverse colon. The two less common types are total colon aganglionosis with extension into small bowel and ultrashort segment which involves 3-4 cm of anal sphincter only.

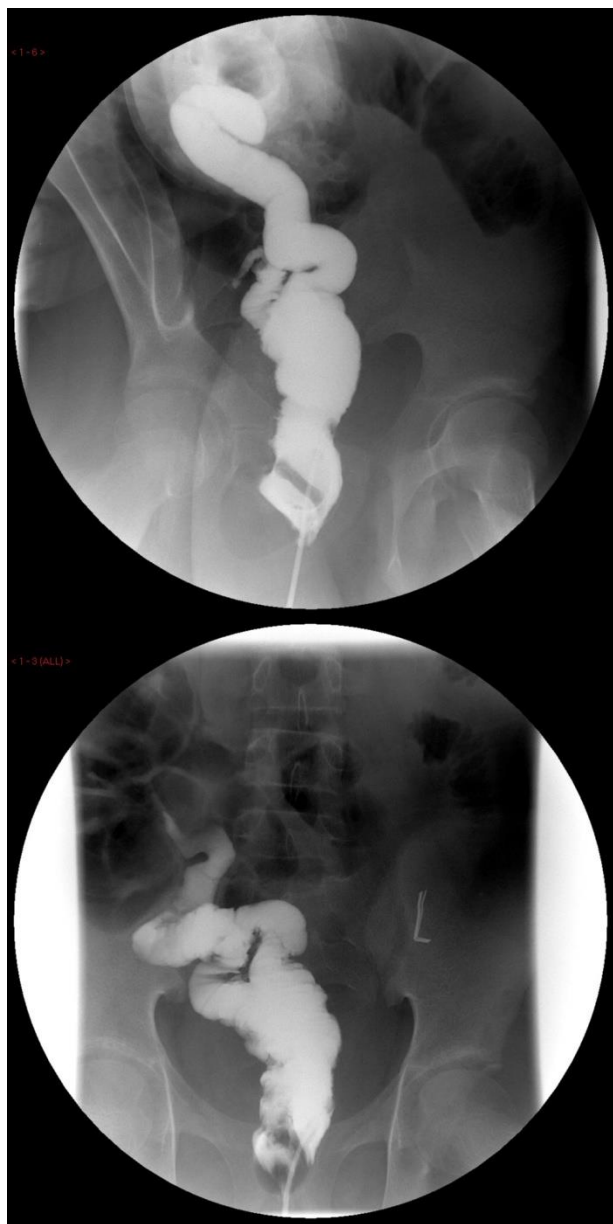


Figure 5: Fluoroscopy Gastrografin Enema, oblique and antero-posterior views, showing well opacified ileoanal pouch with contrast reaching up to ileostomy site, without any contrast leakage or fistulous communication.

Diagnosis of Hirschsprung's disease is incorporates 3 main investigations; the barium enema, anorectal manometry and rectal biopsy. The initial investigation

is barium enema is not only helpful in early diagnosis but also helps to tell the length of the affected segment yet is deficient in exact depiction of the transition between ganglionic and aganglionic segment. It affected segment appears narrow in caliber with proximally dilated normal colonic segment. Saw tooth irregularity and flocculation as also sometimes seen in the affected segment.

Conventional radiography though a good depicter of complications like bowel obstruction showing dilated bowel loops on plain radiograph, plays no role in diagnosis the disease. No definite feature is noted on plain radiograph apart from some mottle lucencies suggested fecal matter in the large bowel due to prolonged constipation.

Computed tomography not a frequently performed investigation for the diagnosis of Hirschsprung's disease plays an important role in milder disease/ long standing case where chronic constipation is the only symptoms. It shows features similar to barium enema with narrow calibered affected segment and dilated normal colon however with better anatomic delineation.

The next investigation performed after barium enema is anorectal manometry which shows failure of the anal sphincter to relax on rectal distention. Followed by the gold standard investigation i.e., the rectal biopsy from the narrowed segment which shows absence of ganglion cells, hyperplasia, and hypertrophy of nerve fibers, and an increased level of the enzyme acetylcholinesterase.

The treatment for Hirschsprung's disease is the surgical removal of the affected bowel loop, with good prognosis in most of the cases.

CONCLUSION

Hirschsprung's disease is congenital disorder; its adult presentation though rare but still an encountered entity. It is a treatable condition provided prompt diagnosis and early surgical treatment is provided. It is important to consider HD in young patients presenting with chronic constipation and failure to thrive. In addition, it is important to anticipate HD in patients presenting with complications of HD like bowel obstruction. Never the less early diagnosis and management cannot only cause enlivening effects on a person's health but can have a major impact on an individual as a whole, by decreasing morbidity.

ETHICAL APPROVAL

The study was approved from Institutional Review Board of Lahore Medical and Dental College/ Ghurki trust teaching hospital, Lahore, Lahore, Pakistan, vide reference No. IRB 140-960-2020, dated April 28, 2020.

REFERENCES

1. Chen F, Winston III JH, Jain SK, Frankel WL. Hirschsprung's disease in a young adult: report of a case and review of the literature. *Annals of diagnostic pathology*. 2006 Dec 1;10(6):347-351.
2. Qiu JF, Shi YJ, Hu L, Fang L, Wang HF, Zhang MC. Adult Hirschsprung's disease: report of four cases. *International journal of clinical and experimental pathology*. 2013;6(8):1624.
3. Iwashita T, Kruger GM, Pardal R, Kiel MJ, Morrison SJ. Hirschsprung disease is linked to defects in neural crest stem cell function. *Science*. 2003 Aug 15;301(5635):972-976.
4. Devos AS, Buonomo C. Radiological imaging of the digestive tract in infants and children. Blickman JG, editor. Springer-Verlag Berlin Heidelberg; 2008.
5. Pescatori M. Benign anorectal disease: diagnosis with endoanal and endorectal ultrasound and new treatment options. *Techniques in Coloproctology*. 2007 Mar 1;11(1):70.

AUTHOR'S CONTRIBUTIONS

SG: Concept and Design, Data collection, Data analysis, Manuscript writing

ZM: Manuscript writing, Drafting

KHB: Critical Review, Literature review