17 years Old Female Hirschsprung·s Disease –a Case report

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Abstract— Adult Hirschsprung's disease (HD) is an uncommon disorder, since congenital aganglionosis is most often diagnosed in infancy and early childhood and present with intestinal obstruction heralded by failure to pass meconium within the first 24 hours of life .However, this case may be present at age of 17 as a result of misdiagnosis or late presentation. We present a case of a 17-year old female who presented with a history of constipation, abdominal pain and marked abdominal distention that improved eventually with conservative management. These intermittent attacks of partial bowel obstruction were treated conservatively since childhood. Developmental millstone was normal and there is no significant family history of Hirschsprung's disease. Diagnosis was made with full thickness rectal biopsy. The patient had functional colostomy for 3 months. After that she underwent Souave endorectal pull-through and followed with incontinence and diarrhea for the first few days that resolved spontaneously with time

Index Terms— Adolescence Hirschsprung's disease, Chronic constipation, Rectal biopsy, Souave endorectal pull-through

1 INTRODUCTION

HD is a developmental disorder characterized by the absence of ganglion cells from the myenteric and sub mucosal plexuses of part of the large bowel results in a narrow, contracted segment.
HD is commonly recognized in the newborn and infants. However, a few may be missed until adolescent period or even late adulthood (1).
Presentation is usually in the neonatal period with intestinal obstruction heralded by failure to pass meconium within the first 24 hours of life. Abdominal distension and later bile-stained vomiting develop. Occasionally, infants present with severe, life-threatening Hirschsprung enterocolitis during the first few weeks of life. In later childhood, presentation is with chronic constipation, usually profound, and associated with abdominal distension but usually without soiling. Growth failure may also present (2).
Diagnosis is confirmed when there is aganglionosis on H&E staining of a rectal biopsy. Staged surgical resection and subsequent re-anastomosis provide good outcome (1).

Case Report:

This is a case of a 17-year old female who presented to emergency department of King Abdulaziz Specialist Hospital, Taif, Saudi Arabia with 13 days ago history of constipation, abdominal pain and marked abdominal distention that improved eventually with conservative management. These intermittent attacks of partial bowel obstruction were treated conservatively since childhood. The patient was not diagnosed histologically as having aganglionosis (Hirschsprung's disease) since then. Developmental milestones had been normal. No similar condition was found in her family. The patient was vitally stable, her chest was normal and she had abdominal distention with visible peristalsis. CT abdomen and pelvis plain with contrast show diffuse marked distention of all colonic segments with no transition zone. By surgical section CT and per rectal examination there was ultrashort segment of rectum constricted. She underwent 3 stages operation. Exploratory laparotomy with colostomy was arranged. Biopsy from proximal and distal part of the colon was taken. The third biopsy was taken from the distal rectal and upper anal canal (full thickness). She had a left sided divided transverse colostomy. Hypoganglionosis at rectal segment was proved by histopathology. She had functional colostomy for 3 months. After that she underwent Souave endorectal pull-through and followed by incontinence and diarrhea for the first few days that resolved spontaneously with time

Discussion:

Adult HD is rare and often undiagnosed or misdiagnosed (3). The first described case of adult HD was reported by Rosin et al. in 1950 (4). According to the detailed study carried out by Miyamoto et al., males are more diagnosed with adult HD than females with a ratio of 133 to 42. Age of HD in adult range between 10 – 73 years with mean 24.1 years (5).
Adult HD presented with chronic constipation, abdominal distention and pain. Regular use of cathartics is required in the majority of patients to relieve their symptoms. Abdominal distention and tenderness are commonly seen on physical examination of the patient. Radiological study of the abdomen displays massive distention of the proximal region of the colon, with a small narrowed distal segment. CT is required to diagnose the disease by showing the dilated colon and the transitional zone and to exclude other diseases with similar presentation like colorectal cancer. History and barium enema are important to diagnose the disease but for definitive diagnosis full-thickness rectal biopsy is essential (6). However, in this case the late presentation might be due to hypoganglionosis in the histopathology. There is no disease with hypoganglionosis so its variant of HD.
In this case, we faced different controversial issues. First, is it better for patient to do colectomy with permanent ileostomy or ileal pouch anastomosis or preserve the colon in our pa-

TABLE 1
UNITS FOR MAGENTIC PROPERTIES

tient? Eventually, we preserve the colon. Patient developed colitis due to the prolonged dilatation that leaded to stasis but it improved with time. Second, is closure of colostomy in separate operation in order to protect recto anal anastomosis a better choice? Then, we do bowel preparation by mechanical and chemical methods. The patient then developed diarrhea and incontinence to fluid. The external sphincter, pudendal nerve and internal sphincter are intact so we were confident that it will be transient and improved by pelvic floor exercise. The last issue was when we had the CT, the entire colon was dilated and there were strictures below the colostomy that either due to skipped lesion of hypoganglionosis. However, when we did the CT in first time, there was no strictures. So, these strictures might be due to cutting in the blood supply but it will lead to gangrene not stricture, or these might be functional structures which was the most reliable explanation.
In study of 229 cases of adult HD, it was found that Duhamel procedure is operation of choice as result of the lower postoperative morbidity rate and better functional outcome (5).
Conclusions:
Chronic constipation, abdominal distention and pain in adult have variety differential diagnosis. Adult Hirschsprung's disease should be suspected when other diseases with similar presentation are excluded. Diagnosis is made by full-thickness rectal biopsy. Patients can be treated with endorectal pull through procedure

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Figures

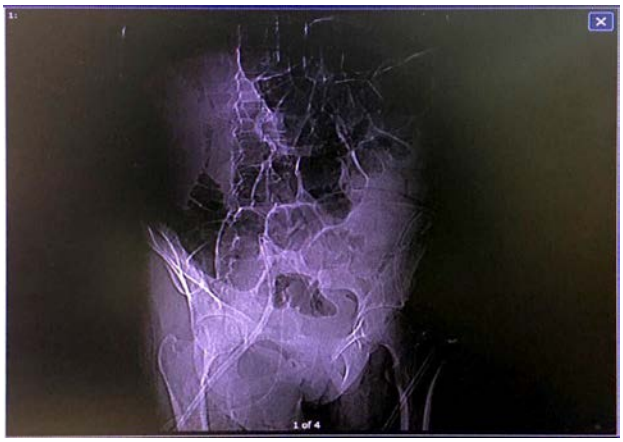
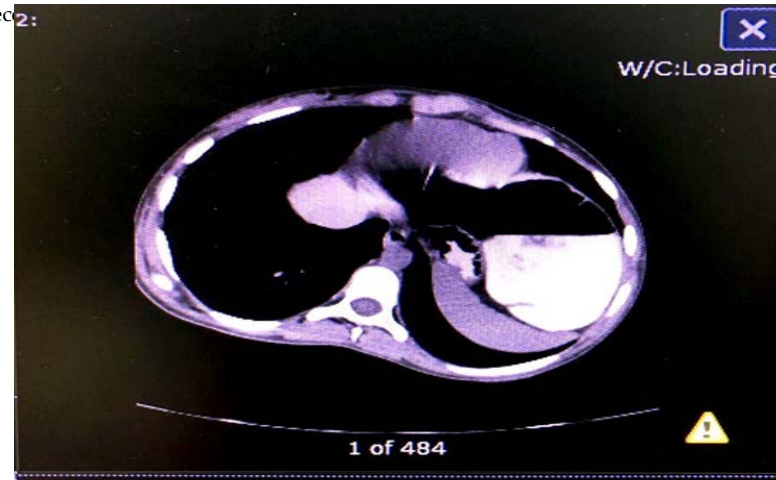
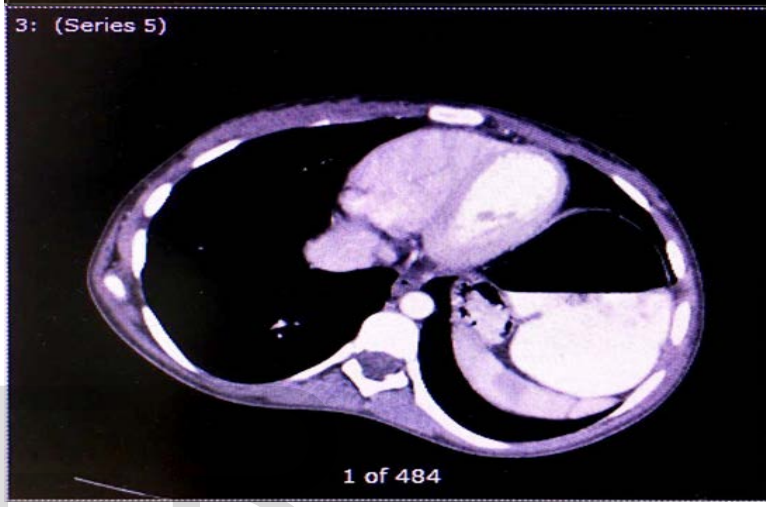
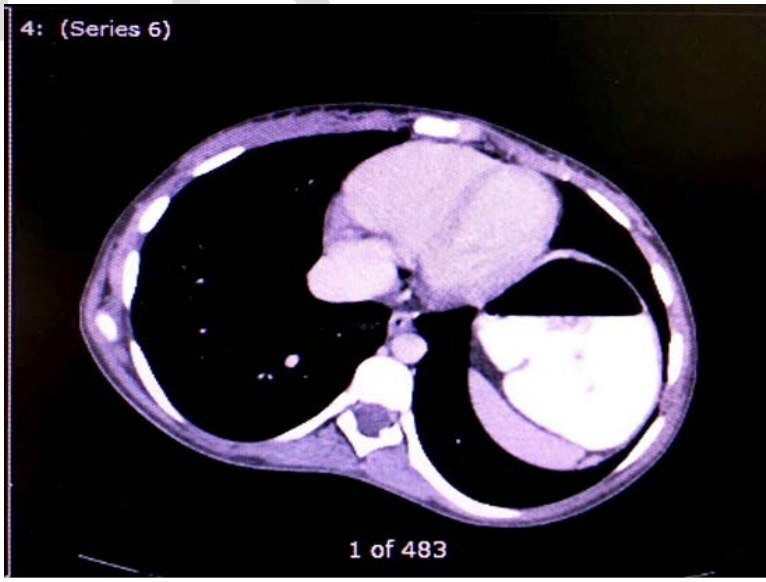


Fig. 1. >>>>>>>>>







Statements that serve as captions for the entire table do not need footnote letters.
[a]Gaussian units are the same as cgs emu for magnetostatics; Mx = maxwell, G = gauss, Oe = oersted; Wb = weber, V = volt, s = second, T = tesla, m = meter, A = ampere, J = joule, kg = kilogram, H = henry.

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