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Peutz-Jegher's Syndrome Presenting as Multiple Intussusceptions: A Case Report.

KEYWORDS: Intussusception, Peutz-jegher's, Enteroscopy, Colonoscopy, Biopsy

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ABSTRACT *The Peutz-Jeghers syndrome is an autosomal dominant disorder characterized by hamartomatous polyposis of the gastrointestinal tract, melanin pigmentation of the skin and mucous membranes, and an increased risk for cancer. Polyps are often a cause of invagination and ileus in affected patients. A long-term follow-up is required to prevent invagination and ileus in children and cancer in adults. Here we present a a case of a previously healthy 25-year-old man with complaints of intermittent pain abdomen, vomiting and loose stools which are suggestive of sub acute intestinal obstruction. Oral and buccal pigmentations gave clues to think about peutz-jegher's syndrome. Multiple intussusceptions were noted on CT and MR enterograohy.The diagnosis was further confirmed by enteroscopy followed by colonoscopy and biopsy.*

Introduction

Intussusceptions is defined as the telescoping of one segment of the gastrointestinal tract into an adjacent one, invagination of a proximal segment of bowel (intussusceptum) into the lumen of the adjacent distal segment (intussuscipiens).While intususception is relatively common in the childhood, it is infrequently seen in adults [1]. In 1921, Dr Johannes Peutz described 7 cases of multiple intestinal polyps associated with pigmentation on lips, buccal mucosa and digits in 3 generations of a Dutch family.[2] In 1939 Dr Harold Joseph Jeghers observed similar symptoms and signs in a 14 year old girl admitted to the Boston Hospital. The family described by Jeghers, in literature referred to as the "Harrisburg family", in which 12 cases of the disease were diagnosed. Much later in December 1949, Jeghers and his coworkers, McKusick and Katz, used the name of "Peutz Jeghers syndrome" for the first time. The name was officially accepted in 1954. [3] A incidence of PJS is estimated to be 1 in 25,000 to 280,000 births. Equal in females and males, appears in every race and all ethnic groups.

Case report

A 25 year old hardware engineer came to opd with the complaints of loose stools, pain abdomen and vomiting intermittently since 6 months. 3-4 episodes of diarrhea with each episode lasting for 1-1.5 months. He was passing three to four semisolid stools per day, moderate in quality which was non-greasy and not associated with pus or blood. The bowel movement took place half to one hour after taking food. It was associated with diffuse cramping abdominal pain which was relieved by vomiting and loose stools. Appetite was normal with no significant loss of weight. There was no history of fever, jaundice, muscle or joint pains. No history of any systemic illness.

Physical examination revealed multiple dark, pigmented spots on his lips and buccal mucosa and similar spots on his finger tips and soles of his feet. He revealed that he has been having them since birth and that his uncle also has similar spots. His vitals and other systems examination was unremarkable.

Laboratory investigations revealed hemoglobin of 10gm%, with normal leukocyte count. Routine blood chemistries, serum amylase were normal. Negative for viral markers. Thyroid profile was normal. A complete stool examination was normal and negative for occult blood. An ultrasound abdomen revealed multiple beaded bowel loops in the right para-colic region with evidence of thickening of the ascending colon, hepatic flexure and transverse colon suggesting hypertrophic tuberculosis involving the above sites. The results of an upper GI endoscopy were normal.

A barium meal follow through was done which showed two filling defects in the jejunal loops suggestive of polyps. In the delayed film another filling defect was noticed in the sigmoid region. A contrast enhanced computed tomography (CECT) of abdomen showed circumferential bowel wall thickening with target or pseudo-kidney appearance at right lumbar region (figure :1) suggesting intussusceptions jejuno-jejunal in the left lumbar and ileo-ileal in the right lumbar region. MR enterography was done confirming pseudo-kidney appearance in the region of right para-colic gutter and left lumbar region suggestive of intussusceptions.(figure :2&3)

Enteroscopy was done and a small pedunculated polyp of size 0.5×0.5cm in proximal jejunum was seen, (figure:4) for which polypectomy was done and sent for HPE. On colonoscopy ,small sessile polyps were visualized in mid – transverse colon and sigmoid colon from which biopsy was taken and submitted for HPE .The microscopic examination of sections of jejunal polyp revealed a core of arborizing

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smooth muscle that supported non-neoplastic small bowel mucosa, suggesting a possibility of a non-neoplastic hyper plastic polyp such as a peutz-jegher's polyp.(figure:5&6) The histopathology of sigmoid polyp had a similar appearance suggesting a hyperplastic polyp favoring a peutz-jeghers polyp and the two sections did not suggest any dysplasia.

In the background of multiple pigmented spots on the lips and buccal mucosa and the presence of hyper plastic intestinal polyps, we conclude this as a peutz-jeghers syndrome presenting as intussusceptions.

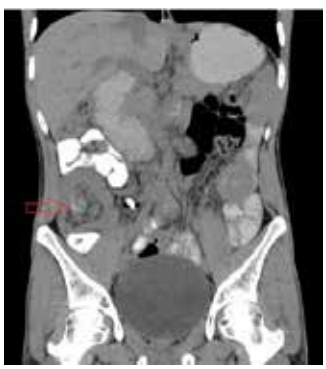
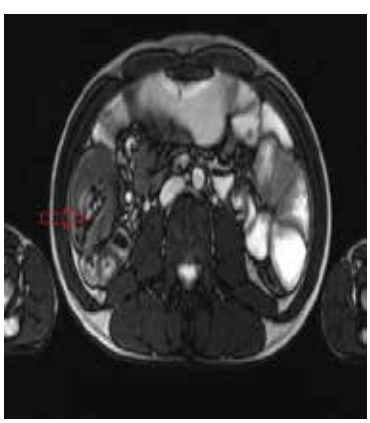


FIGURE-1: CECT-ABDOMEN SHOWING TARGET/ PSEUDO-KIDNEY APPEARANCE (RED ARROW) IN THE RIGHT LUMBAR REGION



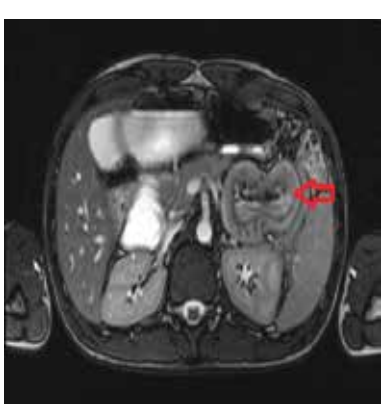


FIGURE-2: MR ENTEROGRAPHY SHOWING PSEUDO-KIDNEY APPEARANCE IN THE REGION OF RIGHT PARA-COLIC GUTTER AND LEFT LUMBAR REGION SUGGESTIVE OF INTUSSUSCEPTIONS

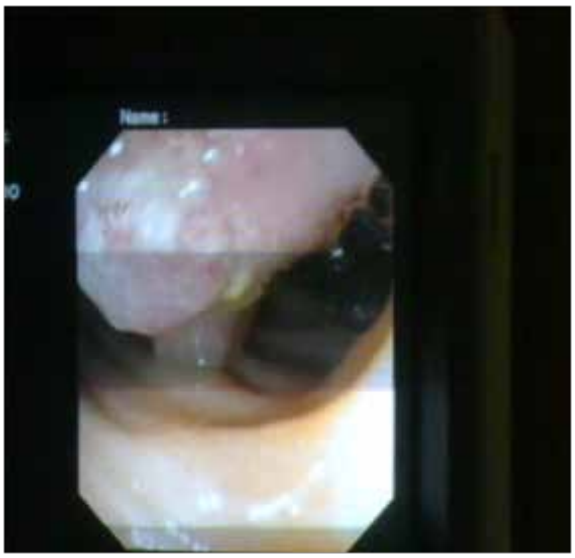


FIGURE-3: ENTEROSCOPIC APPEARANCE OF THE LARGE JEJUNA POLYP.

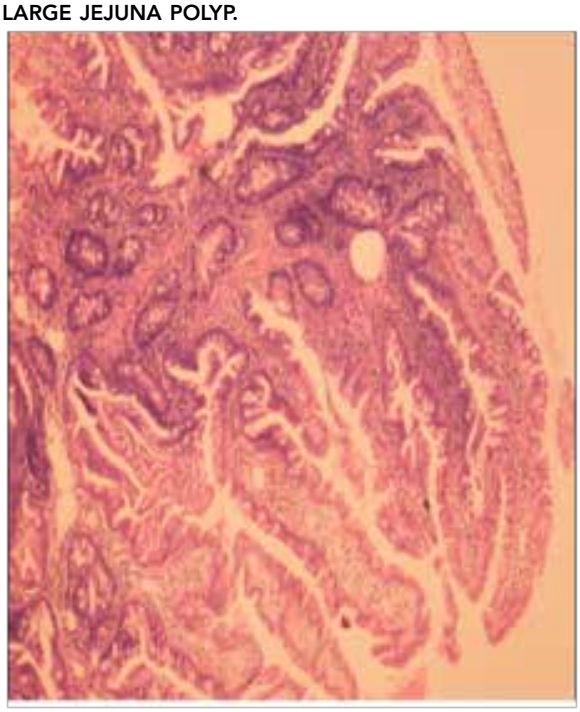


Figure-4: MICROSCOPIC APPEARANCE of sigmoid polyp containing a core of arborizing (branch like) smooth muscle (thick arrow), upon which rests mucosa of small bowel mucosa.gland shows mucinous cells with basally situated nuclei.no adenomatous changes or inflammation .muscularis mucosa appears thickened with mild splaying of fibers.

Discussion :

Idiopathic intussusception although common in children but is less common in adults[4]. In adults, definite causes either Meckel's diverticulum, tumors or polyp will result into intussusceptions [5].

The diagnostic criteria for Peutz-Jeghers syndrome proposed by the Johns Hopkins Registry include histopathologically verified hamartomatous polyps with at least 2 of the following: small-bowel location for polyposis, mucocu-

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taneous melanotic pigmentation, and a family history of Peutz-Jeghers syndrome. The most frequent symptoms are recurrent colicky abdominal pain caused by obstruction and transient intussusceptions. The patient with Peutz-Jeghers syndrome (PJS) should be educated on the potential symptoms of intestinal obstruction and instructed on the need for cancer surveillance.

Periodic surveillance and removal of larger polyps aims to reduce the likelihood of complications in Peutz-Jeghers syndrome. Hence, surveillance for gastric and small-bowel polyposis should begin at age 8-10 years and continue at 2-year intervals. When small bowel polyps are present, there is broad consensus amongst quaternary referral centers that they be removed before symptoms and obstruction become evade.

The diagnosis of intususception may be delayed in adults because of a varied presentation ,although presenting as intestinal obstruction with pain abdomen and loose stools should suspect the intususception [6].

Management includes laprotomy in cases of large bowel intussusceptions but reduction without resection may be a possibility in case of small bowel intusuceptions [7].

Consent

Written informed consent was obtained for publication of this case report and accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal.

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