Peutz-Jeghers Syndrome: A Case Report

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Abstract:

Hamartomatous polyps in GIT are found in all patients of PJ, most commonly the jejunum. One third of patients have symptoms before the age of 10 years and one half before the age of 20 years. Patients most commonly present with obstruction or abdominal pain related intussusception induced by polyps. Intestinal bleeding from the polyps is the second most common GI symptom. Treatment for serious bleedings and intussusceptions in the small bowel related to PJS is resection. [1, 2, 5]

Introduction:

A combination of hamartomatous polyps of smooth muscles and mucocutaneous pigmentations was described by Hutchinson in 1896. The familial hamartomatous polyps was described originally by Peutz and later followed by Jeghers and colleagues. [4] It is an autosomal dominant inherited disorder most commonly resulting from truncating mutations in a serine threonine kinase gene on chromosome 19q (STRK11-LKB1). [10] According to John Hopkins registry, the diagnosis requires two of the following:

- Small bowel polyposis.
- Mucocutaneous pigmentation.
- A family history suggesting autosomal dominant inheritance. [1, 2, 3, 8]

Report of case

An 18 years old Libyan female, presented to the surgical ER with colicky abdomen pain associated with vomiting, the pain started 2 weeks prior to presentation. Episodes of recurrent abdominal pain over the course of 4 years with multiple hospitalizations were reported in her past medical history.Initial examination revealed a dark-brown pigmented maculae on perioral peri-nasal and peri-ocular skin Figure A. A mildly distended abdomen, Soft lax upon palpation, diffuse tenderness and guarding upon central palpation (paraumbilical area). Bowel sounds present. Digital rectal examination reported an empty rectum. Work up for the patient included blood works and radiology. Her CBC elicited marked anemia with a HGB level of 7

mg/dl. Her biochemistry reports sodium and potassium levels of 134, 3.9 Mmol/L respectively. Radiology: Erect abdomen x-ray reveals centrally located dilated bowel loop, Figure B. Further assess with abdomen CT scan with Contrast which illustrated segment of small intestine, mesenteric fat and blood vessels invaginate into adjoining intestinal lumen causing bowel obstruction and giving target sign of intussusception just anterior to left kidney Figure C. Urgent laparotomy was of merit to relive the obstruction, and upon exploration, the bowel health at the intussuscepted segment mandated removal of the segment. So, Resection and Primary Reanastomosis was done. Figure D. Patient recuperated quickly and discharged on the 6th postoperative day.

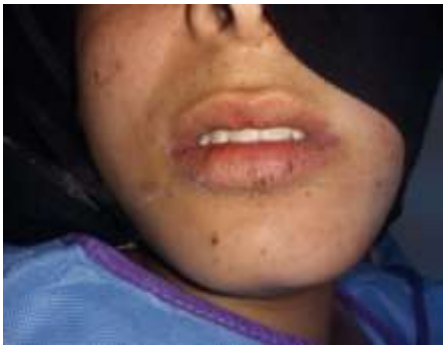

Figure A

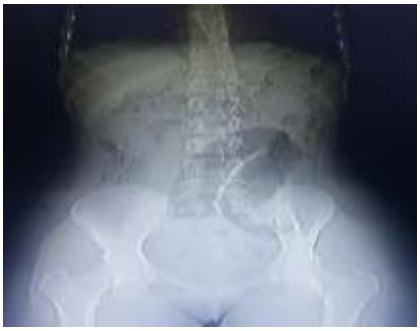

Figure B (Abdomen X-ray dilated Bowel

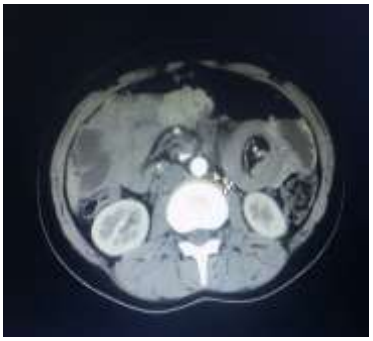

Figure C (revealed target sign of intussesception

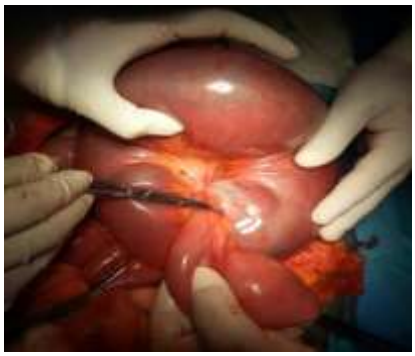

Figure D (site of small bowel intussesception)

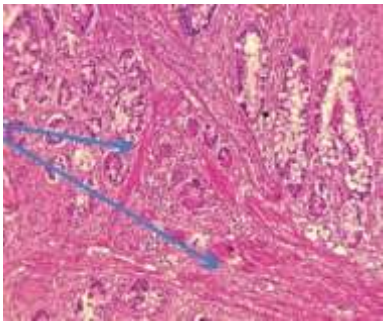

Figure D

Discussion

To stand on solid ground a criteria made by john Hopkins registry was followed to confirm the provisional diagnosis, which requires the presence of the following:

1. Histologically verified hamartomatous polyps.
2. With 2 of the following:
    - Polyp at small bowel.
    - Melanotic pigmentation.
    - A FH of (PJS). [2, 7, 8]

The criteria were met when the histopathology reported the intussuscepted segment of the small intestine has two infarcted polypoid lesions consistent with Peutz Jeghers polyps, the polypoid lesions composed mainly of extensive infarcted intestinal tissue with few preserved area of epithelium and lamina propria intervened by arborizing fascicles of smooth muscles **FigureD**, the histopathology reports absence of any malignant changes.

Conclusion:

To minimize relaparotomies and avoid complications of repeated resections and also halt any malignant growth related to the aforementioned, affected people are advised to undergo regular upper endoscopy, colonoscopy, and small bowel and pancreatic imaging. Polyps > 1 cm in size should be removed as a precautionary measure to prevent complications such as anemia related to chronic GIT bleeding and obstructions due to polyps and intussusceptions that might eventually result in complications such as short bowel syndrome or malnutrition due to repeated resections of recurrent intussusceptions. [6]

Consent

Written informed consent was obtained from the patient for publication of this case report and any accompanying images.

References:-

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