Peutz-Jeghers Syndrome – A Case Report

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Abstract:
Peutz-Jeghers syndrome (PJS) is an autosomal dominant polyposis syndrome and its prevalence is 1 in 100000 individuals. It can be asymptomatic or cause intussusception. Here we present a case of PJS to create awareness and make it a differential diagnosis while treating patients.

Introduction:
Peutz-Jeghers syndrome (PJS) is an autosomal dominant polyposis syndrome. It is characterized by pigmented mucocutaneous lesions and gastrointestinal hamartomatous polyps.

The prevalence of PJS is variable and is calculated to be 1 in 8300 to 1 in 280000 individuals. PJS is probably due to germline mutation in STK11 (LKB1) gene. Peutz first reported PJS in the year 1921. Jeghers, Mc-Kusick, and Katz in the year 1949 described the cases in detail.

WHO's clinicopathological criteria for PJS is:

1. Three or more polyps with histological characteristics consistent with PJS.
2. Any number of PJS-related polyps with a family history of suggestive of PJS.
3. Typical mucocutaneous pigmentation with a family history suggestive of PJS.
4. Any number of PJS-related polyps with characteristic mucocutaneous pigmentation.

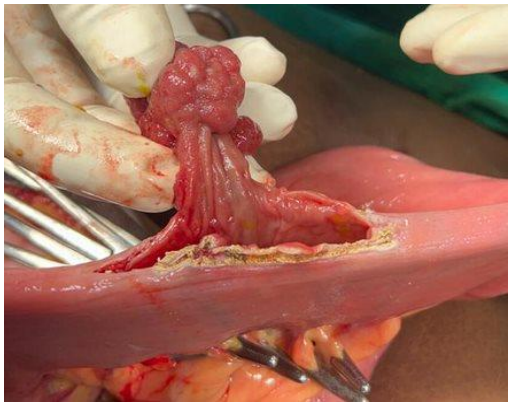
The clinical course of PJS can vary from being asymptomatic to having symptoms like abdominal pain which may progress to intussusception, bleeding, intestinal obstruction, and anaemia. PJS is usually characterized by hyperpigmented mucocutaneous macules present on the perioral region, lips, buccal mucosa, periorbital region, and eyelids. This syndrome also has hamartomatous gastrointestinal polyps. These are most commonly found in small intestine followed by stomach and colon; extra gastrointestinal polyps can also occur. PJS was initially considered as a benign condition, but recently its association with various malignancies, including those of the gastrointestinal tract, lung, pancreas, testis, breast, uterus, and ovary have been published. PJS patients commonly experience bleeding, obstruction, and intussusception.

The management of PJS is therefore aggressive. Double balloon enteroscopy (DBE) and polypectomy are now the treatment of choice. A careful screening is important for PJS. The prevalence of PJS is low. Hence it is important to diagnose early and prevent grave complications. It is also important to screen first-degree relatives for PJS.

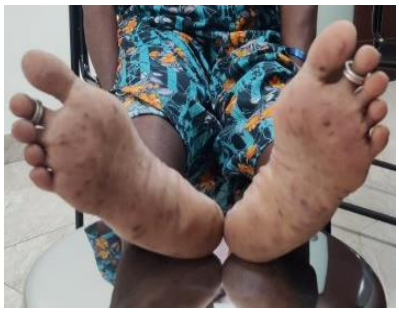
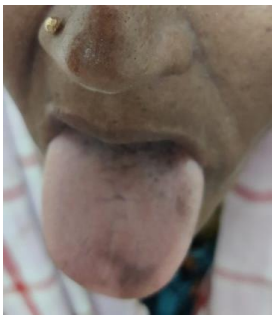
A very few cases of PJS are reported worldwide, hence, we report 53 years old female case with Peutz-Jeghers syndrome.

Case report:
53 years female came with complaints of abdominal distention, pain and vomiting for the past few hours. Her abdomen was rigid, distended and silent, which were suggestive of acute intestinal obstruction. Blood investigations were within normal limits, plain CT whole abdomen revealed small bowel obstruction due to intussusuption.

She was taken up for emergency laparotomy. Intra operatively there were multiple small bowel intussusceptions with intra-luminal polyps. The largest intussuscipiens revealed a 10 cm polyp, 30 cms from the Ileo-cecal junction. Hence that segment of ileum was resected and end to end anastomosis was done. The specimen was sent to histopathology.



Post operatively patient was stable and recovered uneventfully. Multiple skin and mucosal pigmentations were noted in the palms, soles and oral cavity. Patients elder brother had similar plantar pigmentations

Histopathology reported pedunculated and sessile polyps with over lying epithelium in villo-glandular and tubular pattern, without evidence of dysplasia, suggestive of peutz-Jeghers polyposis.

Provisional diagnosis:
Ileal polyp with intussusception

Procedure done:
Laparotomy - ileal resection with anastomosis done.

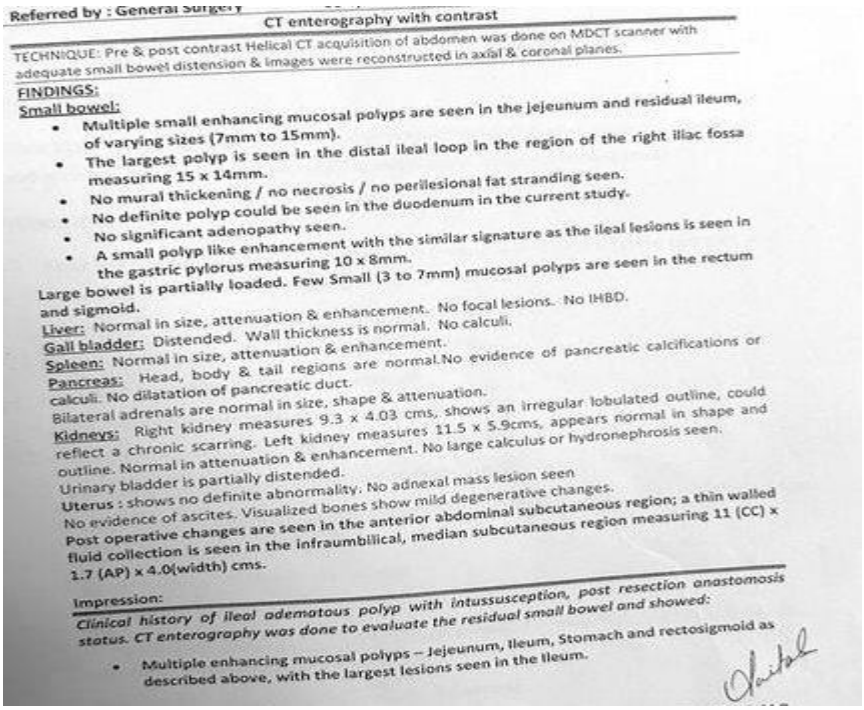
Nature of specimen sent:
Ileum with polyp.

Gross:
Received a segment of small intestine measuring 10cm. External surface- perforated,cut surface -multiple pedunculated and sessile polyps noted (approximately 10) carpetting the ileal mucosa ranging in size from 3.5x2x1.5cm to 0.8x0.5x04cm.Largest polyp is seen protruding through the perforation. Largest polyp is 4.2cm from one resected margin and 6cm from other resected margin. No lymph nodes made out grossly.
Partially embedded.
A,B - Largest polyp
C,D - polyp with Stalk and serosa
E,F,G - Other polyps with serosa
H - Additional bit from largest polyp (without stalk)
I - Distal resected margin
J - Other resected margin

Microscopy:
Sections studied show pedunculated and few sessile polyps composed of arborization of compact smooth muscle bundle. The overlying epithelium is in villoglandular and tubular pattern. No evidence of dysplasia and invasion into the pedicle and the serosa noted.

Impression:
PEUTZ JEGHERS POLYPOSIS.

On POD 14, a contrast CT was done, which revealed multiple enhancing mucosal polyps in the stomach, jejunum, ileum and recto sigmoid. Largest polyp was in the ileum measuring 15x14mm, followed by pyloric polyp which was 10x8mm.



Referred by : General Surgery

CT enterography with contrast

TECHNIQUE: Pre & post contrast Helical CT acquisition of abdomen was done on MDCT scanner with adequate small bowel distension & images were reconstructed in axial & coronal planes.

FINDINGS:

Small bowel:

- Multiple small enhancing mucosal polyps are seen in the jejeunum and residual ileum, of varying sizes (7mm to 15mm).
- The largest polyp is seen in the distal ileal loop in the region of the right iliac fossa measuring 15 x 14mm.
- No mural thickening / no necrosis / no perilesional fat stranding seen.
- No definite polyp could be seen in the duodenum in the current study.
- No significant adenopathy seen.
- A small polyp like enhancement with the similar signature as the ileal lesions is seen in the gastric pylorus measuring 10 x 8mm.

Large bowel is partially loaded. Few Small (3 to 7mm) mucosal polyps are seen in the rectum and sigmoid.

Liver: Normal in size, attenuation & enhancement. No focal lesions. No IHBD.

Gall bladder: Distended. Wall thickness is normal. No calculi.

Spleen: Normal in size, attenuation & enhancement.

Pancreas: Head, body & tail regions are normal.No evidence of pancreatic calcifications or calculi. No dilatation of pancreatic duct.

Bilateral adrenals are normal in size, shape & attenuation.

Kidneys: Right kidney measures 9.3 x 4.03 cms, shows an irregular lobulated outline, could reflect a chronic scarring. Left kidney measures 11.5 x 5.9cms, appears normal in shape and outline. Normal in attenuation & enhancement. No large calculus or hydronephrosis seen.

Urinary bladder is partially distended.

Uterus : shows no definite abnormality. No adnexal mass lesion seen

No evidence of ascites. Visualized bones show mild degenerative changes.

Post operative changes are seen in the anterior abdominal subcutaneous region; a thin walled fluid collection is seen in the infraumbilical, median subcutaneous region measuring 11 (CC) x 1.7 (AP) x 4.0(width) cms.

Impression:

Clinical history of ileal adematous polyp with intussusception, post resection anastomosis status. CT enterography was done to evaluate the residual small bowel and showed:

- Multiple enhancing mucosal polyps – Jejeunum, Ileum, Stomach and rectosigmoid as described above, with the largest lesions seen in the Ileum.

Discussion:

PJS is an autosomal dominant inherited polyposis syndrome, characterised by polyps in the gastrointestinal (GI) tract associated with mucocutaneous pigmentation. PJS is caused by a germline mutation in the STK11 (LKB1) gene.

The diagnosis of PJS can be made in patients with hamartomatous polyp(s) with at least two of the following clinical criteria

1. labial melanin deposits
2. Family history of the syndrome
3. small bowel polyposis

The syndrome occurs equally in males and females and is found in all racial groups. The clinical manifestation of PJS is abdominal pain due to intussusception often leading to intestinal obstruction, polyp extrusion through the rectum, and bleeding.

Small bowel obstruction is the most common presentation in half of these cases. Apart from polyposis, there is an increased risk of GI and extra-GI malignancies. A study showed the relative risk of an individual with PJS to present neoplasia in any region is up to 15 times higher compared to the general population. The most common neoplasm in patients with PJS is the colonic tumor (57%), followed by breast (45%), pancreas (36%), stomach (29%), ovary (21%), small intestine (13%), and uterine tumours (10%). A high risk of pulmonary cancer, renal cancer, prostatic cancer, bone cancer, and leukemia has also been reported.

PJS patients requires special surveillance that includes multiple organs, as it is associated with increased risk of cancer in many organs. Screening begins at 8 to 10 years of age with an evaluation of the small bowel. If initial exam is normal, a repeat evaluation is recommended at 18 years of age , followed by a 2-3 year intervals.

In our case, the patient was admitted to the hospital with intussusception. The exploratory laparotomy revealed intussusception (lead point being polyp). Small bowel resection was performed and end to end ileo-ileal anastomosis done.

Conclusion:

Intussusception is a rare cause of acute abdomen in adults. A high index of suspicion and appropriate investigations can result in prompt diagnosis. 75% of cases are due to malignancies. The extent of resection and operative technique depend upon the age of the patient, results of investigations (benign or malignant) and the length of the bowel involved. Thorough evaluation intra-op and post-op follow up is required in such case to prevent further progression of disease.

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