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Peutz-Jeghers Syndrome Causing Intussusception and Malignancy at an Early Age

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Keywords

Peutz-Jeghers Syndrome, intussusception, bowel obstruction, bowel cancer

INTRODUCTION:

PJS is a rare inherited condition, inherited as an autosomal dominant disorder, due to defect in the serine threonine kinase STK11 gene or LKB1 gene mapped on chromosome 19p13.3. These genes are known for tumor suppression.^[1] The defect in these genes can explain the formation of hamartomatous polyps and predisposition to malignancy.^[2] Genetic tests are used but a negative genetic test does not rule out PJS. A 22 year old gentleman presented with an acute abdomen, and was incidentally found to be having mucocutaneous pigmentation on examination. During the exploratory laparotomy, he was found to be having a jejuno-jejunal intussusception along with multiple polyps, the histopathological examination revealed adenocarcinoma.

CASE PRESENTATION (BODY TEXT):

A 22 year old gentleman presented with complaints of acute severe abdominal pain and multiple episodes of bilious vomiting since the past 12 hours, associated with complaints of constipation and obstipation since 2 days. He was a diagnosed case of PJS based on the presence of oral mucocutaneous pigmentation typical of PJS (Figure-1) and a prior polyp from the duodenum that had histological features of PJS; he was on surveillance for the same.

On examination, he had a pulse rate of 132/minute, respiratory rate of 32/minute, and a blood pressure of 91/64 mm Hg. His abdomen was minimally distended with severe guarding and rigidity and the rest of the systemic examination was within normal limit. Radiograph of the abdomen showed features of acute intestinal obstruction. His CBC, clotting parameters and RFT were within normal limits. With the above basic information, it was decided to proceed with an emergency laparotomy. Intra operatively, a jejuno-jejunal intussusception was found, which was not reducible and hence the involved 15 cm segment was resected (Figure-2) and a jejuno-jejunal end-to-end anastomosis was performed. Another jejunal segment was found to be having multiple palpable polyps which were obstructing most of the lumen. This segment was resected (Figure-3) as well and end-to-end anastomosis was done.



Figure-1: Mucocutaneous hyperpigmented macules (black circles) typically seen in PJS

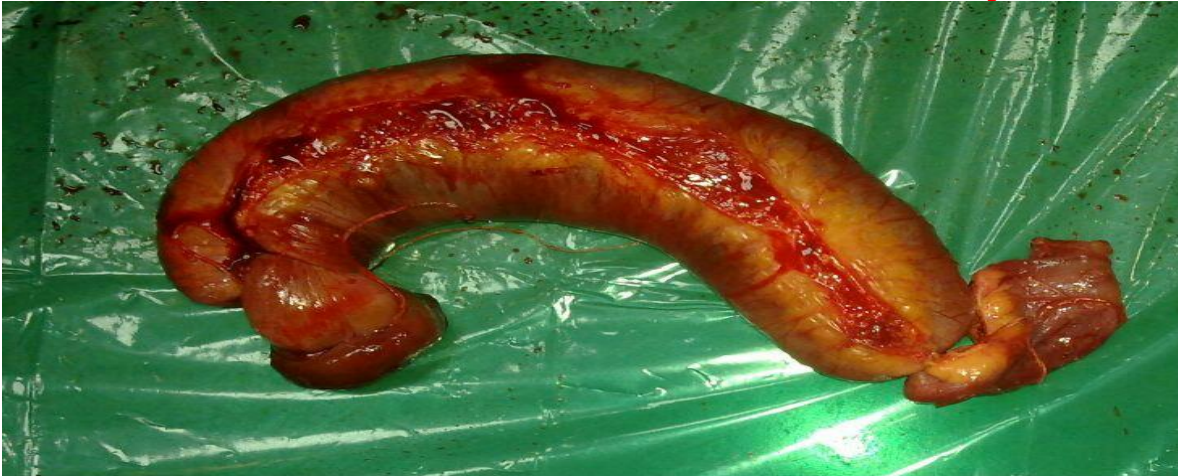


Figure-2: Resected specimen of jejunum-jejunal intussusceptions

The polyps and resected lymph nodes were sent for histopathological examination which showed features suggestive of adenocarcinoma of the small bowel (Figure-4), with the resected margins free of cancer cells; the lymph nodes were negative for cancer cells. His recovery was uneventful. and was discharged on postoperative day 5.



Figure-3: Cut open specimen of same jejunal segment which shows early changes of congestion and polyp leading to irreducible intussusceptions

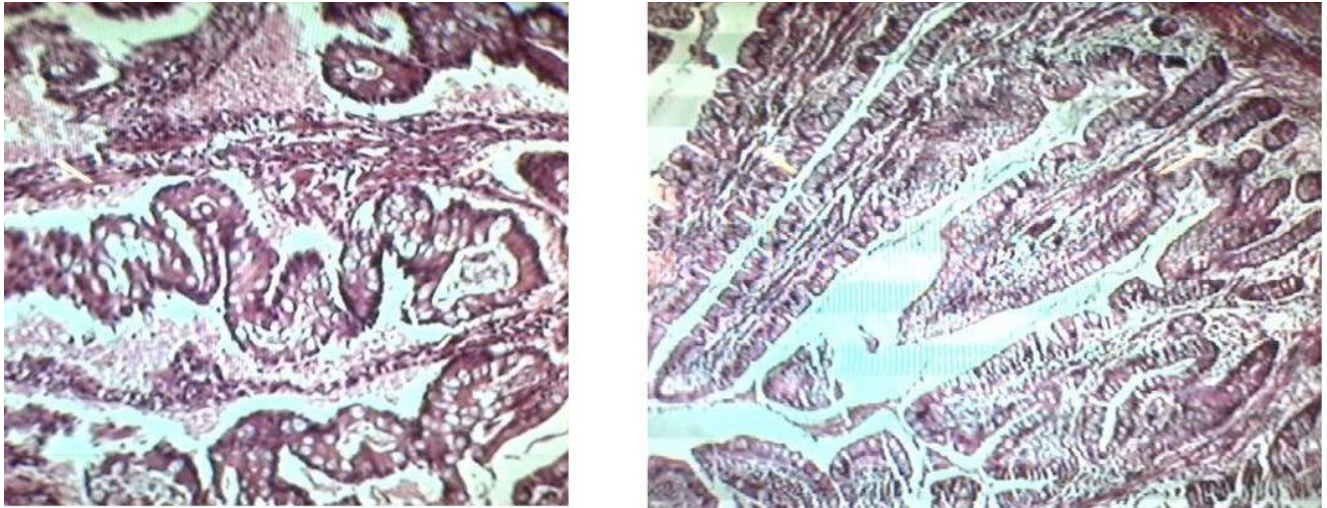


Figure-4: Histopathology showing adenocarcinoma of the resected jejunal segment

DISCUSSION:

PJS is an inheritable, autosomal dominant disorder because of affected STK11 or LKB1 gene which are known for their tumor suppression ability which leads to hamartomatous polyp formation which are highly vulnerable for harboring malignancy^{1,2}.

A family history of polyps, presence of mucocutaneous melanin spots and small bowel polyps makes the likely diagnosis of PJS. Small intestinal polyps are more common mostly in jejunum (>90%) as compared to colonic (64%) and other sites like stomach (49%) and rectum (29%)^{3,4}.

Half of the patients are diagnosed during family screening and the remaining patients present with symptoms such as intestinal obstruction caused by intussusception or direct obstruction of intestinal lumen by the polyp (43%), abdominal pain (23%), gastrointestinal bleeding (14%), or polyp relapsed(7%)^{3,4}.



According to WHO ⁵, the diagnosis of PJS requires the following criteria should be used:

- Positive family history in an individual, with any number of PJS polyps confirmed on histology.
- 3 or more than 3 PJS polyps confirmed on histology with negative family history.
- Prominent mucocutaneous pigmentations which are characteristic of PJS with positive family history.
- PJS polyps with prominent mucocutaneous pigmentations which are characteristic of PJS

As per Hearle N et al, the median age of intussusception is 15 years ⁶. These observations make familial surveillance important because more than 90% patient needs surgical management ^{6,7}.

The risk of malignancy in PJS patients as compared to the general population is 37-93% lifetime risk and relative risk of 10-18 ⁷. Risk of all type of cancer in 20 years of age is around 1% which jumps up to the average of 81% by 70years of age ⁷.

Our patient presented with intussusception leading to intestinal obstruction with development of adenocarcinoma at a very early age (22 Years) which is rare in the natural history of PJS.

A polyp with a median size of 15 mm can produce intussusceptions, hence a polyp of more than or equal to 10 mm to 15mm in surveillance is subjected to polypectomy ⁸. Many attempts to manage patients with conservative approach have been made. For small intestine, balloon assisted enteroscopy (BAE) can be successfully used as it has added therapeutic advantage (biopsy and polypectomy) as compared to capsule endoscopy and radiography ⁸.

Surgical management is needed in intussusception with acute obstruction and malignancy in PJS which are associated with morbidities and have potential to hamper quality of life. Also there is a constant risk of developing short bowel syndrome in patients undergoing re-surgeries ⁸.



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A 'Clean sweep' of small intestines for polyps is attempted by intraoperative endoscopy⁹. Oncel M et al showed that patients who underwent laparotomy along with 'clean sweep' approach had reduced re-laparotomy rates in follow-up⁹.

With recommended plan of surveillance tailored for individual patient and use of BAE can conservatively reduce chances of re-surgeries and associated complications⁸. For surveillance, the most commonly recommended investigations are^{5,10}:

1. OGD (Oesophagogastroduodenoscopy) bi-annually (OGD is done at age of 8 years or early if symptomatic and if asymptomatic can be repeated at 18 years and then every 3 years)
2. Capsule endoscopy bi-annually (can be used in post bowel resection patients).
3. Colonoscopy (started at the age of 50 years and done every 1 to 2 years)
4. Contrast radiography (computerised tomography enteroclysis or magnetic resonance enteroclysis)
(Starting at the age of 12 to 18 years, and then repeated every 2 to 5 years).

Hence, we present a rare case of intussusception and malignant polyp in very early course of PJS. Close surveillance, BAE and 'clean sweep' approach are high yielding in management of PJS and has potential to reduce associated risk of short bowel syndrome, morbidity and mortality and can detect malignancy at an early stage.

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