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Case Report

A rare case of Peutz Jeghers syndrome associated with gastric adenocarcinoma and multiple intestinal polyps

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ABSTRACT

Peutz-Jeghers syndrome (PJS) is a rare hereditary disease, inherited as autosomal dominant fashion (variable penetrance). It is characterized by hyperpigmentation over oral cavity and lips and gastrointestinal hamartomatous polyps. It has incidence rate between 1 in 50,000 and 1 in 200,000 people and prevalence of 1 in 100,000 people. Mucocutaneous pigmentations are most common clinical features in these patients. PJS patients have increased risk of gastrointestinal (more commonly) and extra intestinal malignancies. Incidence rate of gastric malignancy in PJS is around 5%. We presented a rare complication of PJS in terms of gastric adenocarcinoma. A 24 year old female presented with left lower abdominal pain on and off since one year. She had nausea, multiple episodes of gastric vomiting and obstipation since two days. On examination, hyperpigmented patches were present over both lips, had tachycardia. Abdomen was distended and tender. Abdominal ultrasound showed possibility of intussusception and CECT abdomen confirmed long segmental intussusception involving jejunal loops with lead point being polyp/hamartoma and presence of hamartoma/polyp in stomach and jejunum. Patient underwent Exploratory Laparotomy. Jejuno-jejunal intussusception was found. Intussusception could be reduced three big sessile polyps were found in it. Resection with jejuno-jejunal anastomosis was done. Postoperative course was uneventful. Upper GI scopy was done which showed multiple polyps in stomach, one large ulcerated polyp, biopsy from that polyp showed moderately differentiated adenocarcinoma. As the development of malignancies in PJS hamartomas is rare, periodic surveillance in patients and their family members is of utmost importance.

Keywords: Intussusception, Hamartomatous polyp, Peutz-Jeghers syndrome, Intestinal obstruction, Gastric adenocarcinoma

INTRODUCTION

Peutz-Jeghers Syndrome (PJS) is a rare hereditary condition that presents with dark pigmented spots around the mouth and lips, alongside hamartomatous polyps scattered throughout the gastrointestinal tract. It is an autosomal dominant disease with variable penetrance, has an estimated incidence rate between 1 in 50,000 and 1 in 200,000 people and prevalence of 1 in 100,000 people.[1] Individuals with incomplete PJS have only mucocutaneous pigmentation, while others have hamartomatous polyps in the gastrointestinal tract along with mucocutaneous pigmentation. Peutz introduced the condition in 1921, and Jeghers expanded on it with studies in 1944 and 1949.[2] It has male:female ratio of 1:1. The condition can commonly be recognized in childhood or early adulthood. Mucocutaneous pigmentations are most common among the clinical features in these patients. Among the common complications are bleeding, bowel obstruction, and intussusception. There is an

increased risk of both gastrointestinal tract as well as extra intestinal organs; although gastrointestinal are more common. There is a low malignant potential of Hamartomatous polyps, but PJS patients are at increased risk of acquiring malignancies.[3,4]

Clinical diagnosis of PJS can be made if a patient presents with any gastrointestinal polyp consistent with PJS with/without mucocutaneous pigmentation and has family history positive for PJS. There have been found germline mutations in the serine threonine kinase 11 gene (STK11 or LKB1 gene) which is responsible for PJS.[5] STK11, a gene responsible for tumor suppression, is located on chromosome 19p13.3. Laboratory tests for finding STK 11 mutation detection are helpful in diagnosing PJS, but there is no relation between its presence and malignant transformation risk.[6,7]

PJS is an uncommon hereditary disorder, which may stay undiscovered for a long time. A less common feature of PJS is the occurrence of intestinal obstruction due to intussusception, which is often linked to intestinal polyps. Early diagnosis of PJS in patients and concomitant screening of family members can lead to better outcomes.

CASE REPORT

A 24 years old female presented with complaints of on and off left lower abdominal pain since one year. She had developed nausea, multiple episodes of gastric vomiting and absolute obstipation since two days. There was no history of abdominal fullness, tuberculosis or other comorbidities. There was no family history of any gastrointestinal diseases. On examination, the abdomen was mildly distended and tender. Hyperpigmented patches were present over both lips. The patient was vitally stable.

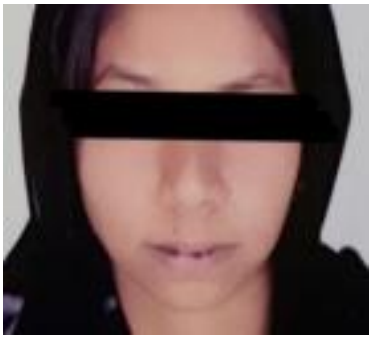

Figure 1: Clinical picture.

Ultrasound of the abdomen showed evidence of bowel within bowel appearance for a long segment (approximately 12 cm) of small bowel loops in right lumbar and right iliac fossa region with possibility of intussusception. CECT abdomen confirmed the diagnosis of intussusception with jejunal loops giving long segmental bowel within the bowel appearance (target sign) in right iliac fossa and right lumbar region for segment of 14 cm; intussusceptum being jejunal loop, mesenteric fat, and mesenteric vessels with few homogeneously enhancing polypoidal soft tissue density lesions. Lead point was largest homogeneously enhancing polypoidal soft tissue density lesion within the intussusceptum measuring approximately 35×27 mm; possibility of hamartoma/polyp. Fluid and content filled prominent jejunal loop were noted proximal to intussusception. CECT abdomen also showed presence of gastric polyps, the largest measuring approximately 40×20 mm.

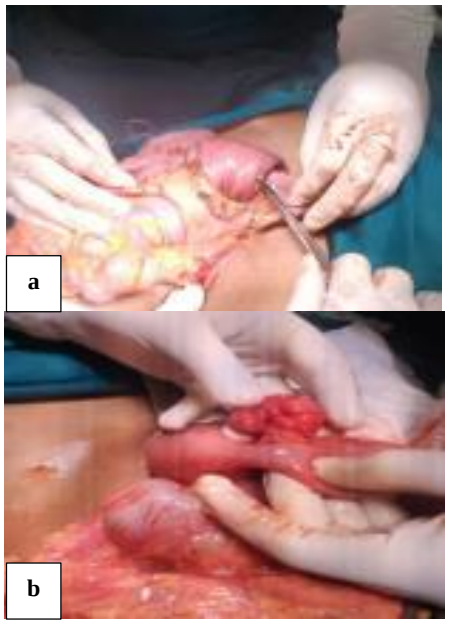

Figure 2 (a and b): Intraoperative image showing intussusception and polyp.

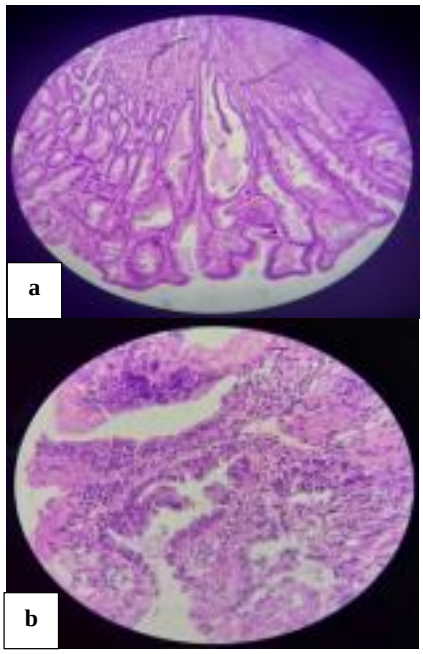

Figure 3: Histopathological image showing (a) hamartomatous polyp (b) gastric adenocarcoinoma.

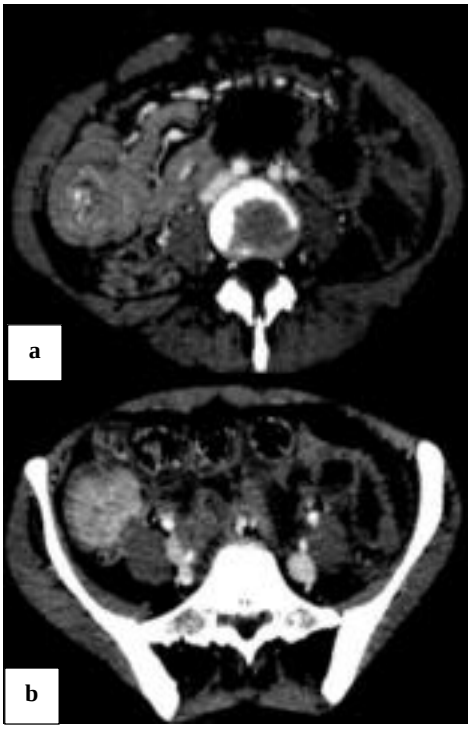

Figure 4: CECT image showing (a) gastric and intestinal hamartomatous polyp (b) intestinal intussusception with hamartomatous polyp.

Patient underwent exploratory laparotomy in view of small bowel obstruction. Approx ten cm long jejuno jejunal intussusception was found about forty cm distal to DJ junction, three big sessile polyps were found in the intussusceptum. Resection of jejunal loops with jejuno-jejunal anastomosis was done. Post operative period of the patient was uneventful. Oral feeds were started on the fourth postoperative day. Histopathology report of excised jejunal loop with polyp was consistent with hamartomatous polyposis (Peutz Jegher's Type).

In view of the presence of gastric polyps, upper gastrointestinal scopy was done on the sixth postoperative day. It showed presence of multiple gastric hamartomas with one large ulcerated polyp. Punch biopsy was taken from ulcerated polyp, which revealed the presence of gastric adenocarcinoma. Patient also underwent a colonoscopy. No polyps were found in rectum and colon.

Patient was discharged on the seventh postoperative day. She was advised metastatic workup and further treatment accordingly for gastric adenocarcinoma. But the patient is lost to follow up.

DISCUSSION

PJS is a rare syndrome which is inherited in autosomal dominant manner and has variable penetrance. It is estimated that worldwide PJS has an incidence of about 1 in 50,000 to 2,00,000 individuals and prevalence of 1 in 100,000 people.[1] It has male:female ratio of 1:1. Usually can be diagnosed during childhood or in some cases in early adulthood.[3,4] Individuals with incomplete PJS only exhibit mucocutaneous pigmentation, while those with the full condition also have hamartomatous polyps in the gastrointestinal tract along with mucocutaneous lentiginosis. Clinical diagnosis of PJS can be made if a patient presents with any gastrointestinal polyp consistent with PJS with/without mucocutaneous pigmentation and has family history positive for PJS. There have been found germline mutations in the serine threonine kinase 11 gene, a tumor suppressor gene, located on chromosome 19p13.3 (STK11 or LKB1 gene) responsible for PJS. Laboratory tests for finding STK 11 mutation detection are helpful in diagnosing PJS, but there is no relation between its presence and malignant transformation risk.[5]

Colorectal cancer (CRC) is the most common cancer in PJS. Breast, small bowel, gastric, pancreatic, and lungs cancers can also be found in these pateints.[8,9] In PJS patients, lifetime risk of malignancy development till the age of 60-70 is 37 to 93% (lifetime cumulative risk of 93%), while for general population is from 9.9 to 18%.[8] Among PJS patients aged 27 to 71 years old, Giardiello et al found that there is an overall risk of 39% of developing CRC, mechanisms of which remain unclear and controversial.[10] According to some hypotheses, the hamartoma-adenoma-carcinoma pathway is responsible, whereas according to some others, PJ polyps may actually represent a form of abnormal mucosal prolapse and do not have malignant potential.[11] Gastric adenocarcinoma in patients with PJS is rarely present, there are very few articles supporting their simultaneous presence.[12,13] It may be an accidental finding to find two separate diseases in a patient or there may be dysplasia of hamartomatous gastric polyp into adenocarcinoma.[14,15]

In our case, the patient presented with clinical features of intestinal obstruction and was found to have multiple jejunal hamartomatous polyps, causing jejunojejunal intussusception. Upper GI scopy revealed presence of multiple gastric polyps with one large ulcerated polyp. On lower GI scopy no polyps were found in colon and rectum. Gastric adenocarcinoma was confirmed by histopathological examination of biopsy taken from ulcerated polyps. This supports the hamartoma-adenoma-carcinoma pathway hypothesis and signifies that hamartomas are associated with malignant transformation, making it important to consider hamartomatous polyps as precursors for malignancy and should be removed taking care of adequate margin clearance.[11] Regular screening of family members and active malignancy surveillance for early malignancies may detect disease at earlier stages and thus improve the eventual outcome of disease. For screening for PJS, nowadays endoscopy in the form of upper gastrointestinal scopy and lower gastrointestinal (colonoscopy) scopy have become essential, capsule endoscopy also aids diagnosis, help in histological confirmation of disease. Literature states that polyps that are more than 1-1.5 cm

in diameter size should undergo polypectomy. Small bowel polyps can be resected either by laparotomy or can be removed via Double balloon endoscopy (DBE).[16]

Major morbidity due to this syndrome apart from malignancy is primarily due to small bowel polyps that require enterotomies as polyps are widespread in the intestine. Though PJS is a very rare condition, around 50% of patients develop intestinal obstruction caused by hamartomatous polyps.[17] This is in contrast to abdominal Koch's highly prevalent in developing countries, where 31.52% of patients develop intestinal obstruction.[18] Some patients may present with complications such as acute or chronic anaemia due to blood loss from ulceration of polyp. Intussusception, though rare, usually presents with a history of intermittent abdominal pain and vomiting. Intussusception may reduce spontaneously or can lead to acute bowel obstruction. PJS can be diagnosed using the WHO criteria, with presence of any one of the following criteria: histologically confirmed more than two Peutz-Jeghers polyps; even single Peutz-Jeghers polyp with a family history is positive for syndrome; mucocutaneous pigmentation with a family history is positive for syndrome; Peutz-Jeghers polyp and mucocutaneous pigmentation.[19]

The presence of mucocutaneous pigmentation over lips and oral mucosa is essential for early diagnosis of PJS, which usually starts in childhood, and may fade or disappear later in life.[2] Our patient had hyperpigmentation over lips and oral mucosa, had multiple hamartomatous polyps in jejunum and gastric adenocarcinoma. According to WHO′ s criteria, diagnosis was confirmed. Macroscopic appearance of polyps is not distinctive for diagnosis, there should be villous architecture with arborizing smooth muscle cores histologically. There could be prolapse and peristaltic kneading which leads to epithelial misplacement and may extend into the serosa, thus can mimic a well-differentiated invasive lesion.[6,20]

CONCLUSION

PJS is a rare disease that often remains undiagnosed for many years. Sometimes the presentation is unusual in the form of intestinal obstruction secondary to intussusception. There is an elevated risk of developing malignancies of both gastrointestinal tract as well as extra intestinal organs, making regular survey of patients mandatory. Screening of family members and thus identifying individuals at-risk for developing malignancies associated with PJS will lead to better outcomes. Early diagnosis of PJS in patients and simultaneous screening of family members can improve prognosis. These individuals require close regular surveillance for better prognosis.

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