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CASE REPORT

Peutz-Jeghers Syndrome with Synchronous Adenocarcinoma Arising from Ileal Polyps

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Abstract Peutz-Jeghers syndrome is a rare inherited autosomal disease characterized by mucocutaneous pigmentation and multiple polyps in the gastrointestinal tract. The clinical picture is characterized by repeated episodes of polyp-induced intestinal obstruction, abdominal pain, and bleeding per rectum. Predisposition to both gastrointestinal and nongastrointestinal malignancies is increased in a patient with Peutz-Jeghers syndrome. This is a case report of a 29-year-old male with Peutz-Jeghers syndrome who presented with a complaint of recurrent abdominal pain. CT scan revealed a single obstructing ileal polyp. However, preoperatively, another large ileal polyp with multiple small jejunal polyps was seen. Histopathology of both ileal polyps was identified as a mucinous adenocarcinoma infiltrating up to the serosa. The follow-up endoscopies showed small multiple polyps in the stomach, duodenum, and colon. Histopathology of all endoscopically removed polyps was consistent with the diagnosis of hamartomatous polyps. Very few cases of intestinal intussusception combined with synchronous malignant small intestine polyps have been reported until to date.

Keywords Peutz-Jeghers syndrome · Synchronous ileal cancer · Hamartomatous polyps · Polypectomy

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Introduction

Peutz-Jeghers syndrome is an autosomal dominant condition characterized by the melanin deposition around the mouth, lips, oral mucosa, and multiple gastrointestinal hamartomatous polyps. Polyps are usually found in the small intestine more commonly in the jejunum followed by the ileum and duodenum [1]. Previously, it was believed that there is low incidence of transformation of hamartomatous polyp to malignant polyp. However, evidence increasingly indicates that there is an increased risk of malignancy both in these polyps and in other organs of the body [2]. Peutz-Jeghers syndrome has been clearly associated with increased risk of small intestinal adenocarcinoma [3]. Until now, few cases have been reported with synchronous small intestine malignancy.

Methods

A 29-year-old male presented with complaints of recurrent lower abdominal pain and chronic constipation for last 6 months. He had a history of intermittent hematochezia. There was no history of nausea, vomiting, and abdominal distension. He had undergone appendicectomy in childhood. There was no history of any comorbid illness. On examination, there were melanotic pigment spots at the mucocutaneous junction of lips (Fig. 1), palms of hands, and soles of feet. Per abdomen examination revealed a palpable vague mass in the right lower abdomen and a well-healed scar mark of appendicectomy in the right iliac fossa. Contrast-enhanced CT (CECT) scan abdomen revealed a mass in the right iliac fossa formed by distal ileal intussusception with a possibility of soft tissue density mass within the intussusception. In view of his symptoms and CECT scan findings, he was taken up for laparotomy. Preoperative

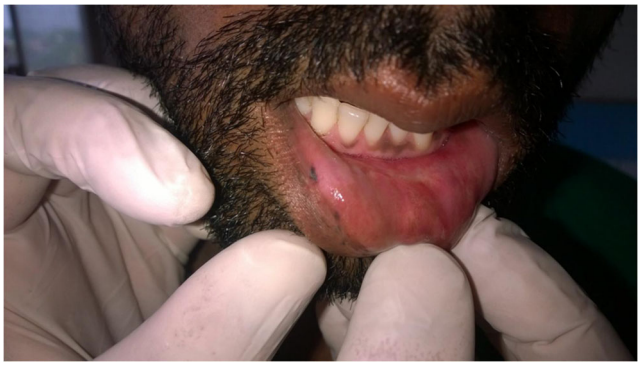

Fig. 1 Melanin pigment spots at the mucocutaneous junction of lips

findings revealed dense adhesions of small bowel and omentum in the right iliac fossa due to previous appendicectomy plus multiple interloop bowel adhesions. A polyp with a size of 3.0×2.5 cm which was obstructive in nature was present about 50 cm from the ileocecal junction. Another large polyp with a size of 2.5×2.0 cm was present in the ileum about 160 cm from the ileocecal junction. In addition, there were multiple small palpable polyps <1.0 cm in the proximal jejunum. Rest of the examination did not reveal any abnormality. He underwent segmental resection of ileal segments bearing polyps followed by end-to-end anastomosis (Fig. 2). His postoperative recovery was uneventful. Histopathology of both the polypoidal lesions was reported as a mucin-secreting adenocarcinoma infiltrating up to the serosa. He received 6 cycles of folinic acid, fluorouracil, and oxaliplatin (FOLFOX) regimen in the postoperative period. He has been on regular follow-up for last 1 year and is asymptomatic.

Follow-up screening with upper gastrointestinal endoscopy revealed multiple polyps <1.0 cm, two on the greater curvature of stomach and one in the third part of duodenum. Endoscopic polypectomy was done. Histopathology of all the polyps was reported as hamartomatous polyps. Colonoscopy also showed multiple pedunculated polyps, one in the proximal transverse colon and two in the sigmoid colon. All the polyps were dealt with endoscopic polypectomy. Histopathology was consistent with the diagnosis of hamartomatous polyps.

Discussion

Peutz-Jeghers syndrome is a rare disease with incidence of one in 8300 to 23,000 live births [4]. It is inherited in an autosomal dominant pattern; however, 50 % of cases could be sporadic and may represent new mutation [5]. Peutz-Jeghers syndrome is associated with germ line mutations in the serine threonine kinase 11 (STK11/LKB1) gene. This gene is localized in the chromosome segment 19p13.3. Serine threonine kinase 11 (STK11/LKB1) gene mutation is found in 30–70 % of sporadic cases of Peutz-Jeghers syndrome and 70 % of affected individuals with a family history of this condition [5]. In the present case, there was no relevant family history.

Polyps in Peutz-Jeghers syndrome are a typical hamartomatous polyp with an arborization pattern of smooth muscle proliferation. The most commonly affected site of polyps in Peutz-Jeghers syndrome is the jejunum, colorectal, duodenum, and stomach in decreasing order. The most common histology type is hamartoma [6]. In our case, multiple hamartomatous polyps were present in the stomach, duodenum, jejunum, and colon. Peutz-Jeghers syndrome is a tumor-susceptible syndrome. It is associated with an increased risk of gastrointestinal and nongastrointestinal malignancies. The incidence of tumor is 15 times the general population, and the incidence of malignancy is up to 20 %. Giardiello in a meta-

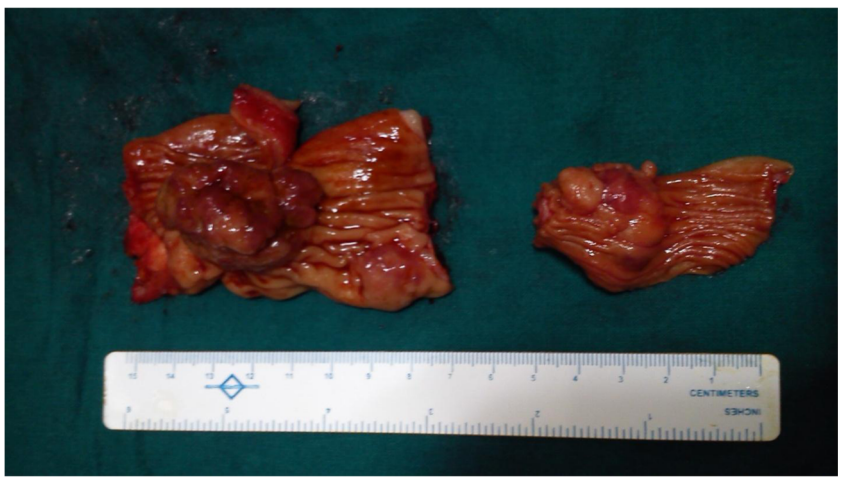

Fig. 2 Resected ileal segments with polypoidal lesions

analytic study has shown a markedly increased lifetime risk of cancer of the colon (39 %), stomach (29 %), intestine (19 %), pancreas (36 %), and esophagus (0.5 %) [6]. There is also an increased lifetime risk of malignancy in the breast (54 %), ovaries (21 %), uterus (9 %), and lung (15 %). Small intestine adenocarcinoma has been detected in a child as young as 13 years of age [7], with patients in the third decade having a cumulative cancer risk of 5 % which rises to 85 % by the age of 70 years.

The present case is in his third decade of life with a synchronous mucinous adenocarcinoma detected in both the ileal polyps. Very few multifocal synchronous cancers in the small intestine have been reported in the literature [8].

This case report describes classical clinicopathological description of Peutz-Jeghers syndrome complicated by synchronous mucinous adenocarcinoma in ileal polyps. It reiterates the fact that Peutz-Jeghers syndrome is a predisposing factor for gastrointestinal cancers.

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