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Peutz Jegher Syndrome presenting as intussusception- A rare case with review of literature

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Abstract

Peutz Jegher syndrome is an autosomal dominant condition characterized by hamartomatous polyps in the intestinal trat with hyperpigmented mucocutaneous macules having greater risk of gastrointestinal and non-gastrointestinal malignancies. The syndrome is rare and infrequently patients present with intussusception. We present an interesting case of Peutz Jegher syndrome in 19 year old male patient presented with intussuception who underwent emergency laproscopic resection of bowel.

Keywords: Petuz Jeghers Syndrome, intestinal polyp, bowel obstruction, secondary malignancies

Introduction

Peutz-Jeghers Syndrome (PJS) is a rare autosomal dominant disorder characterized by hamartomatous polyps in the intestinal tract associated with hyperpigmented mucocutaneous macules seen in 90% of patients(1). The disease affects male and female patients equally. Most patients have a characteristic clinical course of recurrent episodes of polyp induced bowel obstruction and bleeding. These patients have increased risk of intestinal and extraintestinal malignancies(2).

Case Report

A 19 year old male patient presented with pain in abdomen and vomiting since 20 days. The pain was exaggerated since 1 day. X ray abdomen revealed dilated small bowel loops with multiple gas-fluid levels suggestive of small bowel obstruction.

Patient had mucocutaneous pigmentation near medial canthus and on lower lip (Fig 1 & 2)

Emergency laparotomy was done and the resected segments of jejunum and ileum were sent for histopathological examination.

Histopathological Examination

Excised segments of jejunum and segments of ileum were received for histopathological examination.

Segment of jejunum was 78cm in length and showed a dilated part of 25cm with blackish discoloration. On cutting open it revealed multiple pedunculated polyps, largest measuring 2.5 x 1.5 cm. Dilated part of jejunum revealed blackish discoloration and edematous mucosa.

Other two segments of ileum also revealed 2 polyps arising from mucosa measuring 2 x 1.5cm &
1.5 x 1.3 cm (fig 3)

Microscopy of polyps revealed complex arborizing cores with bundles of smooth muscle supporting cystically dilated glands lined by low cuboidal epithelium without nuclear atypia. Dilated portion of the jejunum revealed changes of hemorrhagic necrosis and inflammatory reaction with viable surgical margin (Fig 4)

Based on these clinical and histopathological features, diagnosis was given as Peutz Jeghers polyps with intussusception.

Discussion

Peutz jeghers Syndrome is rare with estimated incidence varying between 1 in 30000 to 1 in 280000 live births [3,4]. The syndrome was initially described by Jan Peutz who noticed a link between mucocutaneous macules and the gastrointestinal polyps in members of Dutch Family [5]. In 1949 Harold Jeghers formalized this into a syndrome[6].

According to World Health Organisation a diagnosis of Petuz Jeghers Syndrome should satisfy any one of the following criteria.

1) Three or more Peutz -Jeghers polyps confirmed by histology.
2) Any numbers of Peutz -Jeghers polyps with a family history of the syndrome.
3) Visible mucocutaneous pigmentation with a family history of the syndrome.
4) Any member of Peutz -Jeghers polyps and visible mucocutaneous pigmentation[7,8].

Our patient presented with multiple Peutz -Jeghers polyps with pigmented macules on lower lip and medial canthus.

Patients with Peutz Jeghers syndrome are at increased risk of acquiring malignancies of colorectal tract, stomach, small intestine, lung, liver, uterus, cervix, ovaries and other organs[7,9]. The lifetime risk for all cancers goes as high as 93% [10]. Gastrointestinal cancers are most common amongst these[11]. Intestinal obstruction is seen in 47-69% of cases, as seen in our case[12]. The management of these cases is laparotomy with resection of bowel loops if polyps are symptomatic or greater than 1.5 cm in diameter. Sizable percentage of patients suffer from short bowel syndrome as a result of repeated bowel resection.(13) Recently intraoperative endoscopy and endoscopic polypectomy have been recommended.

Surveillance protocols for individuals with Peutz Jeghers syndrome has two objectives. One is to identify malignancy at early stage and other is to detect intestinal poylps that could cause obstruction or bleeding. Surveillance include upper endoscopy and colonoscopy.

Conclusion

Peutz Jeghers syndrome is a rare autosomal dominant condition. A high index of suspicion is needed for diagnosis, otherwise patients may present with complications such as bowel obstruction and secondary malignancies. Early diagnosis and close cancer surveillance leads to excellent prognosis.

Legend Figures

Figure 1: Clinical photograph showing pigmentation near medial canthus

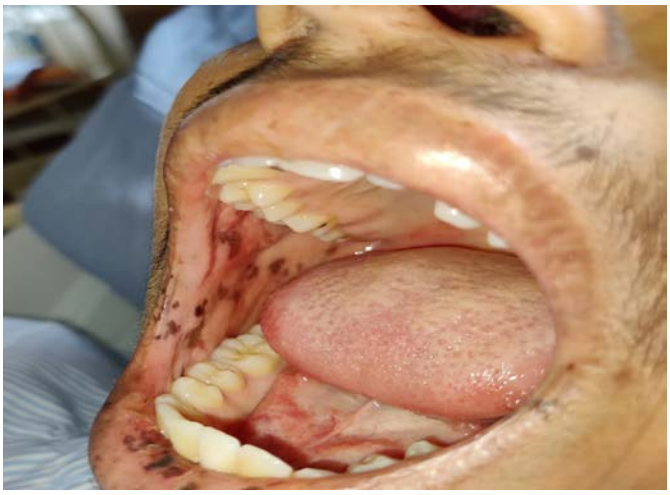

Figure 2: Clinical photograph showing pigmentation on lower lip

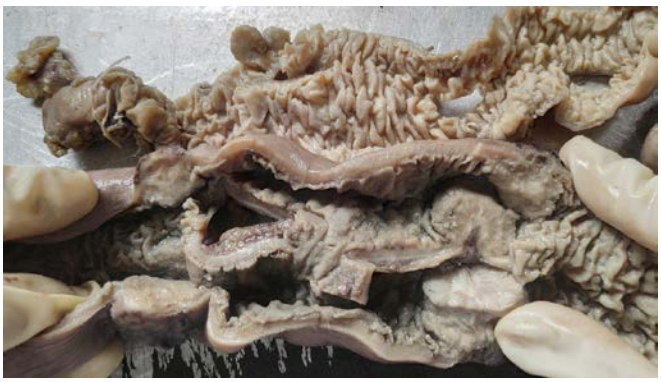

Figure 3: Photograph of resected segment of jejunum & ileum showing multiple polyps

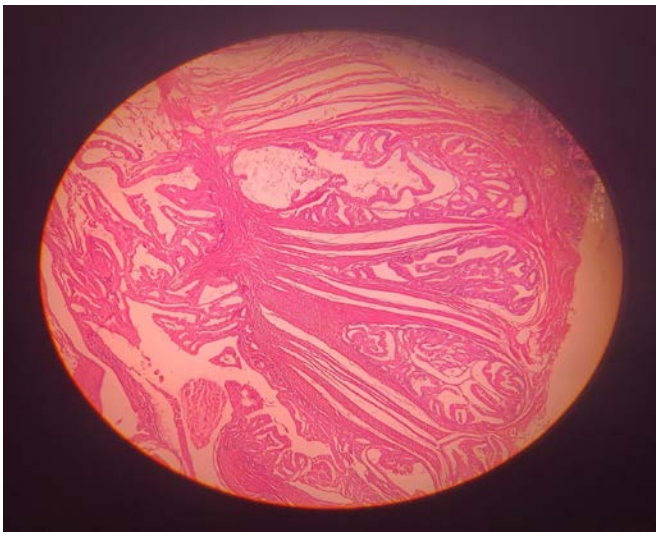

Figure 4a

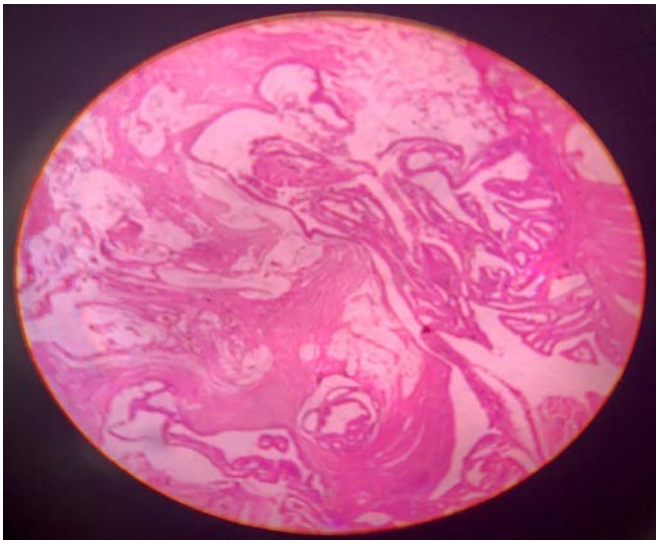

Figure 4b

Microscopy of Peutz Jeghers polyp showing arborising bundles of muscles supporting cystically dilated glands

(a-100x and b-400x)

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