

Rationale for Different Options of Support in Peutz jeghers syndrome

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Abstract

Introduction: Peutz-Jeghers syndrome (PJS) is a rare autosomal dominant disorder characterized by hyperpigmentation on the lips and oral cavity and gastrointestinal (GI) hamartomatous polyps. PJS patients usually present with acute complications such as bleeding, bowel obstruction and intussusceptions.

Case Presentation: We hereby report a case of a 25-year-old male, who presented with complaints of colicky abdominal pain for 4 days. On abdominal examination, tenderness in the right iliac fossa and increased bowel sounds. Patient presented 3 months ago with bleeding per anal and rectal mass. Ultra sound of the abdomen demonstrated suspected ileo-colic intussusceptions. The patient underwent colonoscopy showed pedunculated polyp and polypectomy from ileocaecal region. The symptoms did not subside after 2 days, hence was taken up for a mid-line laparotomy that showed an ileo-colic and jejunojejunal intussusceptions.

Reduction of this intussusceptions was successfully done with, multiple enterotomy and polypectomy. The specimen further confirmed the diagnosis of hamartomatous Peutz-Jeghers polyps with no malignancy. Physical examination revealed multiple pigmented spots on the oral cavity and palm. Thus, the diagnosis of PJS was made.

Discussion: PJS is a rare inherited disease that often remains undiagnosed for many years. Patients often presents with acute complications such as bleeding per rectum, intussusceptions or obstruction.

Conclusion: Early detection and identification of PJS in patients and their family members can improve the prognosis, prevent complications. Patients should be on regular follow up once the diagnosis has been made.

Keywords: Ultra Sound, Intussusceptions, Bleeding, Bowel Obstruction.

Introduction

Peutz-Jeghers syndrome (PJS) is a rare autosomal dominant disorder characterized by hyperpigmentation on the lips and oral cavity and gastrointestinal hamartomatous polyps.¹ It can be diagnosed using clinical criteria which includes the presence of ≥ 2 Peutz-Jeghers polyps anywhere in GI tract, or at least one intestinal Peutz-Jeghers polyp with family history of PJS or classic muco-cutaneous pigmentation.² This disorder is seen uniformly in both genders and not diagnosed until late childhood or early adulthood when they present with some complications.^{3,4} Acute intestinal intussusception is rare in adults, but one of the common complications of PJS. It represents 5 % of all intussusceptions and accounts for 1-5 % of all intestinal obstruction in adults.⁵

Case presentation

A 25-year-old male, who presented with complaints of colicky abdominal pain since 4 days. Her vitals are stable. Physical examination revealed multiple pigmented spots on the oral cavity and palm. On abdominal examination, tenderness in the right iliac fossa and increased bowel sounds.

Patient presented 3 months ago with bleeding per rectum and rectal mass. Patient gives history of resection and anastomotic done in view of gangrenous bowel 10 years ago, was not evaluated further.

Ultra sound of the abdomen demonstrated suspected ileo-colic intussusceptions. The patient underwent colonoscopy showed pedunculated polyp and polypectomy from ileo-caecal region. The symptoms did not subside after 2 days, hence was taken up for a mid-line laparotomy that showed an ileo-colic and jejuno-jejunal intussusceptions. Reduction of this intussusceptions was successfully done with, multiple enterotomy and polypectomy.

The specimen was sent for histopathological evaluation. On gross examination, multiple grey white to grey brown polypoidal tissue.

On microscopy, multiple polyps show polypoidal tissue lined by intestinal epithelium with characteristic arborizing network of connective tissue, smooth muscle, lamina propria and glands, suggesting hamartomatous Peutz-Jeghers polyps with no malignancy. Thus, the diagnosis of PJS was made.



Figure 1: hyperpigmented lesion over the lower lip

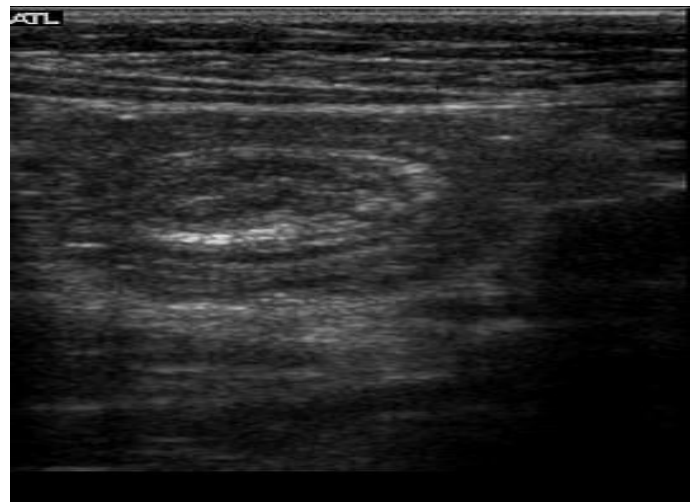


Figure 2: Ultrasound showing ileo-colic intussusception (target sign)

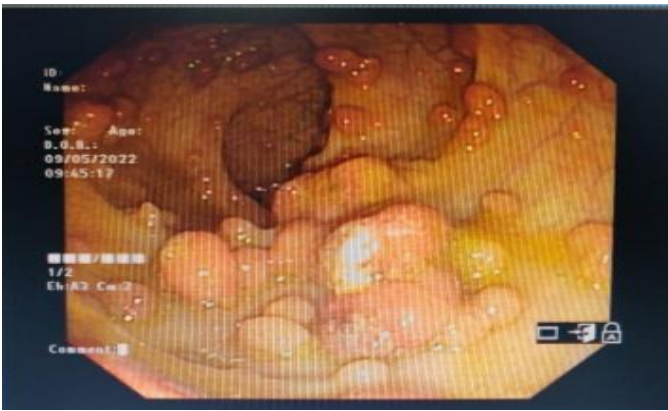


Figure 3: upper GI scopy showing multiple hA Martomatous polyps over the stomach.

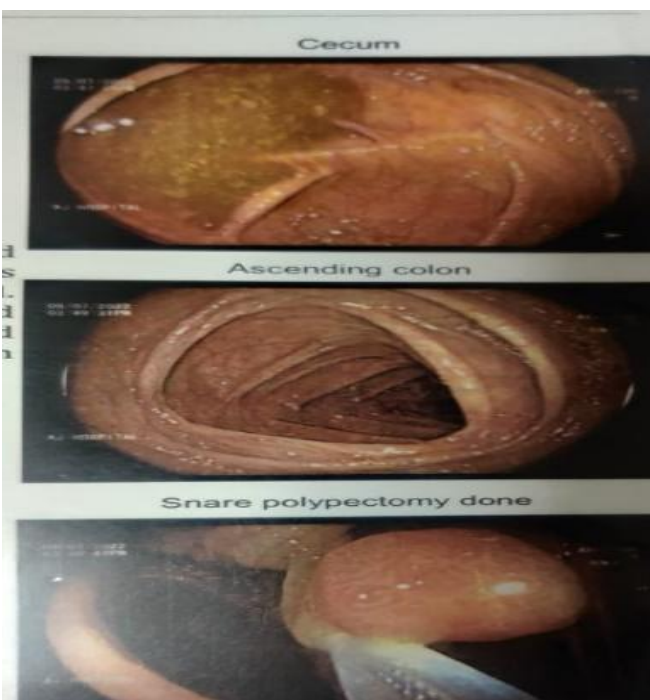


Figure 4: Caecum filled with stool and intussusceptions, snare polypectomy



Figure 5: Ileo-colic intussusceptions

Discussion

PJS is a rare inherited disease that often remain undiagnosed for many years, reported to be 1 in 100,000 individuals⁶. Patients with hamartomatous polyp(s) who also meet at least two of the following clinical criteria can be diagnosed with PJS: small bowel polyposis, labial melanin deposits, and a family history of the syndrome. The syndrome affects all racial groups and affects both men and women equally⁷. Asymptomatic intervals interspersed with consequences including stomach pain, intussusception, which frequently results in intestinal obstruction, polyp extrusion via the rectum and bleeding which is frequently occult, characterize the clinical appearance of PJS. In half of the cases, small intestinal obstruction is the initial complaint, and relaparotomy due to polyp-induced complications happens often and may do so at relatively frequent intervals^{8,9}. Patients usually present to hospital with complications as we see in our case patient presented with pain abdomen and diagnosed to have ileo-colic intussusceptions where the polyps were acting as lead point for intussusceptions. Earlier PJS was considered benign condition and was treated conservatively. Patient was kept on observation and underwent laparotomy when presented with complications, this resulted in multiple laprotomies hence patients are recommended to undergo endoscopy and polypectomy for polyps in the stomach or colon that are greater than 1 cm in size noted during endoscopic surveillance⁹. Our patient underwent colonoscopy which confirmed ileocolic intussusceptions which couldn't be reduced. Patient was posted for exploratory laparotomy, reduction of this intussusceptions was successfully done, bowel segments were found to be viable. Multiple enterotomy and polypectomy carried out to reduce the risk of intussusceptions in the future.

Conclusion

Early detection and identification of PJS in patients and their family members as well as close cancer surveillance can improve the prognosis, prevent complications. Patients should be on regular follow up once the diagnosis has been made.

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