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**Case Report**

# Surveillance in Peutz-Jeghers syndrome: a case report

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## ABSTRACT

Study performed surveillance endoscopy in a 23-year-old male patient with Peutz-Jeghers syndrome (PJS) who underwent right hemicolectomy with ileo-colic anastamosis for ileo-colic intussusception three years back. On evaluation, he found to have multiple upper gastrointestinal and ileal polyps. Subsequently he underwent polypectomies in three sessions by combined standard snare polypectomy and endoscopic mucosal resection (EMR) method without any complication. We present a case report that emphasizes the importance of surveillance and the role of prophylactic polypectomy in patients with PJS.

**Keywords:** Endoscopic mucosal resection, Hamartomatous polyposis, Peutz-Jeghers syndrome, Polypectomy, Surveillance

## INTRODUCTION

Peutz-Jeghers syndrome (PJS) is an inherited, autosomal dominant disorder, characterized by pigmented mucocutaneous lesions and intestinal hamartomatous polyposis. Prevalence of PJS varies from 1 in 8300 to 1 in 280000 individuals. The most common complications associated with PJS are anemia, bleeding, intussusception and intestinal obstruction. Patients with PJS are prone to develop gastrointestinal, pancreatic, breast, lung, uterine, ovarian and testicular tumors. Hence, proper surveillance is essential in this group of patients.[1] Here we report a young patient with PJS, and history of right hemicolectomy with ileo-colic anastamosis who had underwent multiple sessions of endoscopic polypectomy during surveillance.

## CASE REPORT

A 23-year-old male patient with PJS who underwent right hemicolectomy with ileo-colic anastamosis three years ago for ileo-colic intussusception was admitted for surveillance endoscopy. The family history was not significant. On general examination, he had hyperpigmentation in the lips, buccal mucosa and tongue. Physical examination revealed a healthy post laparotomy scar in the midline of the abdomen (Figure 1).

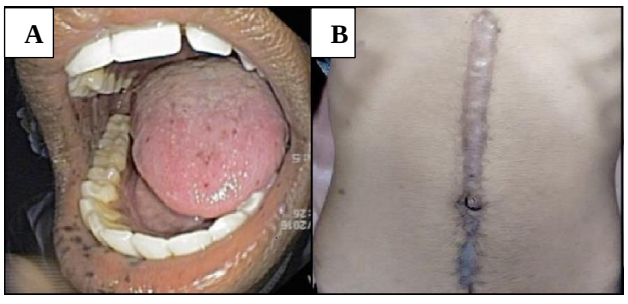


**Figure 1: A) Hyperpigmentation in the lips, buccal mucosa and tongue. B) Healthy post laparotomy scar in the midline of the abdomen.**

Examination of other systems including scrotal examination didn't reveal any abnormality. Laboratory

investigations revealed mild anemia with hemoglobin of 9.1gm/dL. Coagulation profile was normal. Ultrasonography of abdomen was normal. A decision was made to perform esophagogastroduodenoscopy (EGD) and colonoscopy under conscious sedation with simultaneous endoscopic polypectomy for polyps with size of more than 1cm if recognized. The procedure was performed by a highly experienced interventional gastroenterologist. The written informed consent was obtained for the procedure. The EGD showed multiple tiny sessile polyps both in fundus and body of the stomach. In addition, two other sessile polyps were noted in the antrum each measuring 1x1cm. Multiple polyps of varying sizes were also present in first and second part of duodenum (Figure 2). Five pedunculated polyps in duodenum were removed by using traditional snare polypectomy (Figure 3).

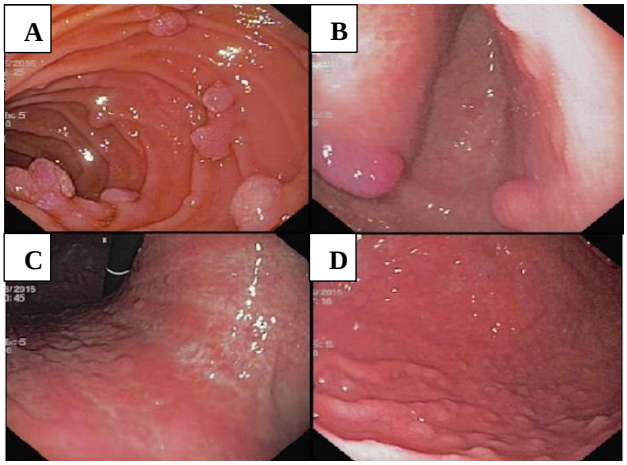


**Figure 2: EGD showing mutiple polyps of varying sizes seen in second part of duodenum (A). Two sessile polyps present in the antrum each measuring 1x1cm (B). Multiple tiny sessile polyps seen in the fundus (C) and body of the stomach (D).**

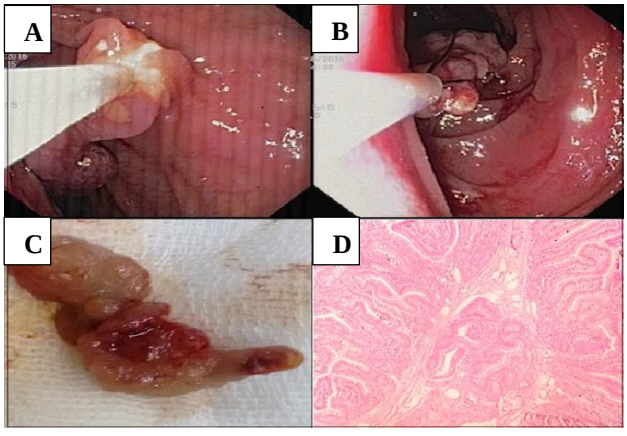


**Figure 3: Pedunculated polyps in duodenum were removed by using traditional snare polypectomy (A, B and C). Histopathology pictures from duodenal polyp, depicts arborizing bands of muscularis mucosa separating mucosal glands which are lined by histologically unremarkable duodenal mucosa, exhibiting Christmas tree pattern (D, H and E 40X).**

In addition we decided to do endoscopic mucosal resection (EMR) of two sessile antral polyps by applying simple injection assisted snare technique. A solution of saline and adrenaline (1 in 20,000) was injected into the submucosal layer to elevate the lesion. After confirming the lifting sign, those two sessile antral polyps were removed en bloc (Figure 4).

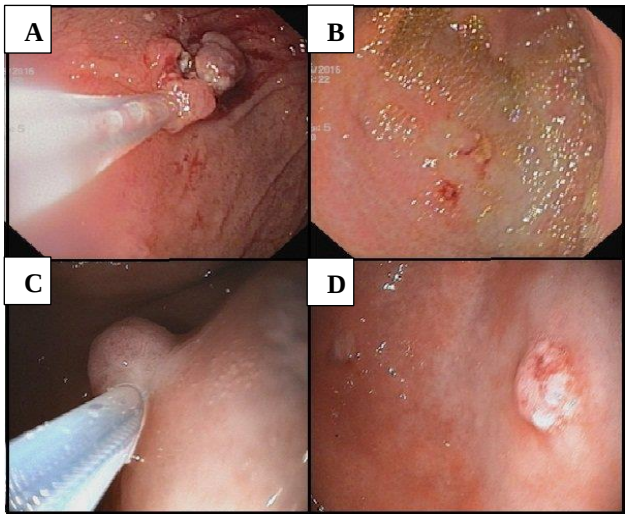


**Figure 4. Endoscopic mucosal resection (EMR) of two sessile antral polyps.**

We couldn't able to remove one pedunculated polyp in duodenum which was not clearly visible in forward viewing endoscopy. Hence two days later, by using side viewing gastroduodenal scope the polyp was removed with standard snare polypectomy. Mild oozing was noted at the polypectomy site, but it stopped spontaneously. After a week, colonoscopy was performed with scope being passed upto 40cm beyond the ileo-colic anastomotic site revealed few tiny sessile polyps and a single large pedunculated polyp beyond ileo-colic anastomotic site (Figure 5). En bloc resection of ileal polyp was done by means of standard cautery polypectomy technique (Figure 6).

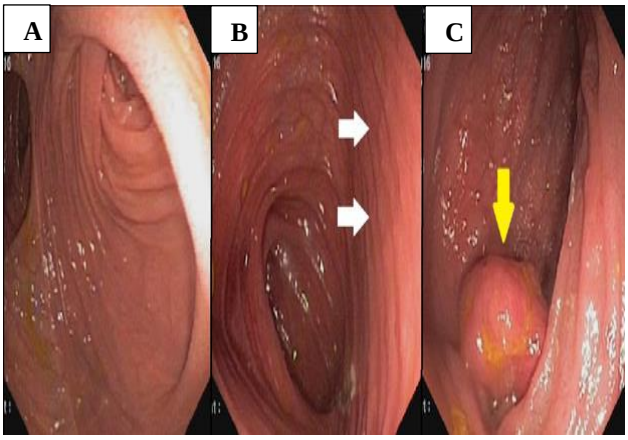


**Figure 5: Colonoscopy shows ileo-colic anastomotic site (A). Few tiny sessile polyps (B) and a single large pedunculated polyp beyond ileo-colic anastomotic site (C).**

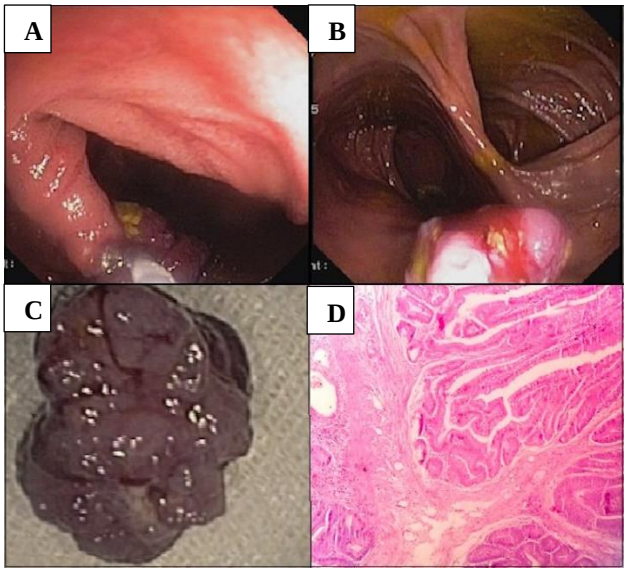


**Figure 6: En bloc resection of ileal polyp was done by means of standard cautery polypectomy technique (A, B and C). Microscopy shows features of ileal polyp with branching muscle fibers dissecting through the mucosal glands. There is no evidence of atypia (D, H and E 40X).**

## DISCUSSION

In patients with PJS the most common identifiable mutation accounting for 60% of cases is the STK11/LKB1 (serine/threonine kinase) mutation, in the locus 13 of the short arm of chromosome 19. Due to the existence of genetic heterogeneity some of the genes are not yet identified.[2] The proposed risk of cancer development at age of 20, 30, 40, 50, 60, and 70 years are estimated as 2%, 5%, 17%, 31%, 60%, and 85% respectively.[3] It is estimated that approximately only 25% of patients with PJS have negative family history.[4]

Study patient also had negative family history. In patients with PJS, polypectomy is recommended if the polyp size is more than 1cm in order to prevent the occurrence of intestinal obstruction.[5] Usually in PJS polyps are most commonly seen in small intestine and colon than stomach.[6] Our patient had polyps in duodenum, ileum and also in stomach. As a total nine polyps were removed (7 pedunculated, 2 sessile polyps) out of which 2 larger polyps were retrieved and sent for histopathological examination. Microscopic examination of the specimen was consistent with PJS (Figure 3D and 6D).

In study patient, out of nine polyps removed and only one associated with minimal oozing of blood which was subsided spontaneously. There were no other immediate or late complications related to polypectomy either by using standard snare polypectomy or applying EMR technique was observed. Combined surgical and intraoperative endoscopic polypectomy will achieve a polyp free intestine, thereby could avoid multiple enterotomies and risk of short bowel syndrome and also allows longer symptom free periods. This modality of treatment is very important in young individuals who will require repeated surgical interventions due to complications of PJS.[7,8] However, it's an invasive procedure and also associated with disadvantages like prolonged post-operative ileus and small bowel leak.[8] On the other hand, the newer tool double balloon enteroscopy (DBE) is less invasive and more compliant for the patients. It is used for both diagnostic and therapeutic purpose. Hence this modality also averts emergency laparotomy and other complications in individuals with PJS.[9,10]

Now with advent of video capsule endoscopy (VCE), DBE and EMR technique the entire small bowel polyps can be removed. So, it's becoming more popular modality of choice worldwide. But this technique is also associated with drawbacks like bleeding and perforation of small intestinal wall. Hence it should be performed by an experienced interventional gastroenterologist in a well-equipped centre. Our patient was advised for further screening of the rest of the bowel using VCE, but patient was not willing for the procedure, thus not performed.

However, he was motivated to undergo the procedure at a later date. Family members screening was also performed and ruled out PJS. We advised regular follow up and emphasized regarding the importance of surveillance to all family members which plays important role in part of the management of this disease. In developing countries like India where patients most often do not turn up for follow up, it is mandatory to do regular counselling for the patient and all family members and moral support too is essential.

The surveillance programme was also suggested to the patient that included, regular screening with EGD, colonoscopy and VCE once in every three years to look for any gastrointestinal malignancy. The patient from the age of 30 years should undergo magnetic resonance cholangiopancreatography or endoscopic ultrasound, once in every 1-2 years to screen for the pancreatic malignancy as per the recent American College of Gastroenterology (ACG) guidelines.[11]

## CONCLUSION

This report highlights the importance of surveillance in patients with PJS and also the use of colonoscopy to screen distal small bowel in patient with post hemicolectomy and ileo-colic anastamosis.

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