

# Peutz-Jegher's syndrome with gut maltoma: A rare presentation

Anindya Mukherjee, Subrata Chattopadhyay, Md. Azam, Aramita Saha<sup>1</sup>

Department of Radiotherapy, Medical College and Hospitals, <sup>1</sup>Department of Medical Oncology, Apollo Gleneagles Hospitals, Kolkata, West Bengal, India

## ABSTRACT

Peutz-Jegher's syndrome (PJS) is an autosomal dominant disorder characterized by numerous hamartomatous polyps in the gastrointestinal tract (GIT) and pigmented muco-cutaneous lesions. We present here a case of a maltoma associated with multiple hamartomatous polyps detected in a post-operative ileocolic specimen of a 28-year-old man. Prior to this, he had undergone surgery for intussusception when similar polyps were noted in the small bowel. Upper GIT endoscopy also confirmed the diffuse presence of such polyps. A clinico-pathological diagnosis of PJS was made, which by itself is rarely encountered. Furthermore, the detection of mucosa associated lymphoid tissue lymphoma/maltoma in a background of PJS is remarkably unique in this case, for which it has been reported.

**Key words:** Hamartomatous polyps, maltoma, Peutz-Jegher's syndrome

## INTRODUCTION

The widest estimated population prevalence of Peutz-Jegher's syndrome (PJS) varies from 1 in 8300 to 1 in 280,000 individuals.<sup>[1,2]</sup> Nearly half of the patients experience an attack of intussusception in their lifetime, mostly due to small bowel polyps.<sup>[3]</sup> Characteristic mucocutaneous macules are observed in 95% of cases around mouth, nostrils, distal extremities, etc., Although PJS is commonly associated with many gastrointestinal and extra intestinal malignancies, lymphoid degeneration on hamartomatous polyps consistent with maltoma has not been reported previously, to the best of our knowledge. Traditionally, resection of only symptomatic polyps has been recommended as standard therapy,<sup>[3]</sup> which was followed in our case.

## CASE REPORT

A 28-year-old man presented to our out-patient department with the complaints of repeated episodes of pain abdomen,

vomiting and constipation over the last 8 months. He reported that his father suffered from similar symptoms too. In March 2013, he presented at emergency with acute abdominal pain and vomiting for which an exploratory laparotomy was performed and numerous polyps giving rise to intussusceptions were discovered in the small intestine. The gangrenous gut was resected out and the polyps were histopathologically found to be benign in nature, consistent with hamartomas. Endoscopic biopsy of polyps detected in D1 and D2 segments of duodenum showed similar polyps and colonoscopy revealed a large polyp in upper ascending colon obstructing the lumen significantly [Figure 1]. An elective right hemicolectomy was done next month. The pathological report described the polypoidal mass to be composed of hyperplastic mucous glands of different size with no infiltration into submucosa or beyond it. Furthermore destructive lymphoid proliferation with characteristic polypoid cells on the surface infiltrating to germinal center was noted [Figure 2]. The histopathological report was suggestive of mucosa associated lymphoid tissue lymphoma. On immunohistochemistry (IHC), CD 20 was found to be diffuse and strongly positive while CD5, CD 23, cyclin D1 and B-cell lymphoma 2 were all negative. All the surgical margins were reportedly negative for malignancy.

## DISCUSSION

PJS belongs to a heterogeneous group of hamartomatous polyposis syndromes which are inherited in an autosomal dominant fashion.

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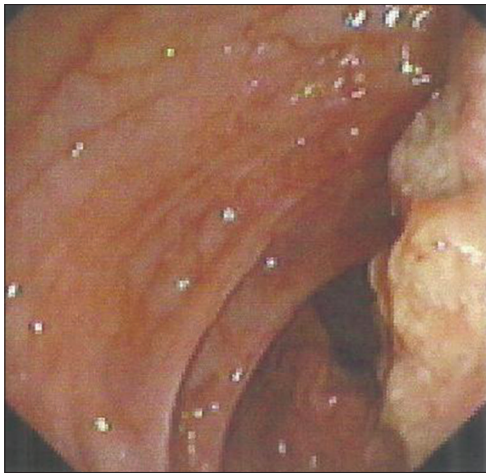
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**Address for correspondence:** Dr. Anindya Mukherjee, Department of Radiotherapy, Medical College and Hospitals, 88, College Street, Kolkata - 700 073, West Bengal, India. E- mail: aniradonc@gmail.com



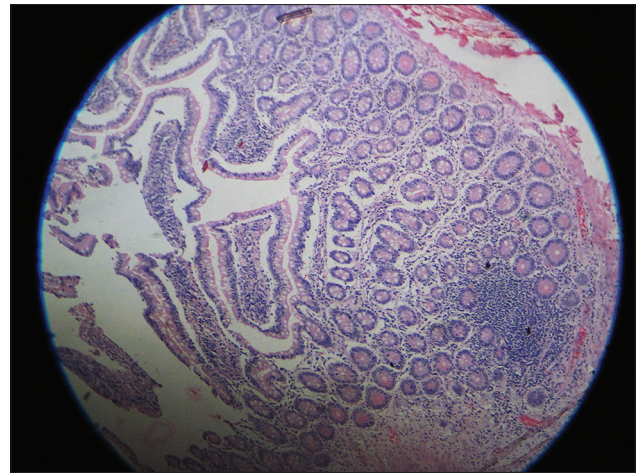
**Figure 1:** Polyp in upper ascending colon

Approximately 50% of patients experience symptoms due to polyps by 20 years of age. Colicky abdominal pain, rectal bleeding, obstruction/intussusceptions are common presentations within first three decades.<sup>[1,3]</sup> Our case corroborates with this typical history of PJS.

Diagnosis of PJS requires the presence of histopathologically confirmed hamartomatous polyps and at least two of the following clinical criteria: Family history, hyperpigmentation and polyps in the small bowel.<sup>[1]</sup> PJS polyps are most commonly observed in small intestine, especially jejunum followed by colon (53%), stomach (49%) and rectum (32%).<sup>[1]</sup> The pigmentations in PJS are typically brown or bluish-gray macules distributed in buccal mucosa, near lips, nostrils and sparsely in palms, sole, intestinal mucosa. Our case is distinguished by the absence of any such pigmentation.

Histologically hamartomatous polyps are characterized by distorted architecture of normal intestinal cellular elements with extensive smooth muscle proliferation and an arborized pattern of polyp formation.<sup>[4]</sup> The presence of a smooth muscle core uniformly arborizing throughout the polyp is the uniquely distinguishing feature of PJS polyps. In our case, destructive lymphoid proliferation was additionally detected and confirmed to be maltoma by IHC, as described above. Confirmations by molecular analysis/cytogenetics were not possible due to lack of this facility in our center. Since it was a stage I non-gastric maltoma resected out with negative surgical margins, follow-up alone was recommended.

Although resection of only those polyps causing recurrent obstruction/intussusceptions has been advocated (which was done in this case), two new modalities in the management of small bowel hamartomas have gained wide acceptance, namely intra-operative endoscopy<sup>[5]</sup> and double-balloon enteroscopy.<sup>[6]</sup> In both these procedures entire small bowel/colon can be visualized and all polyps are removed surgically or endoscopically.



**Figure 2:** Microscopic picture of histopathology showing destructive lymphoid proliferation with characteristic polypoid cells on the surface infiltrating to germinal center

PJS is associated with the specific genetic alteration *STK11/LKB1* mutation and increased risk of developing gastrointestinal and extra-intestinal malignancies,<sup>[7]</sup> especially gall bladder and colonic adenocarcinomas, gynecological and breast malignancies. In a meta-analysis of Giardiello *et al.*,<sup>[8]</sup> the cumulative risk of developing any cancer in PJS was 93%. For this reason, life-long malignancy screening during follow-up has been advocated. Barium enterography, wireless capsule endoscopy, computed tomography (CT)/magnetic resonance imaging enteroclysis with oral contrast, double contrast CT colonography are useful tools to screen gastrointestinal polyps. Routine pelvic examination, chest and abdominal imaging, mammography, etc., can detect extra intestinal malignancies at the earliest.

Numerous case reports of PJS have been reported in the recent past, viz., solitary PJS polyp in jejunum in a 19-year-old male without any other features like multiple polyps or mucocutaneous pigmentation,<sup>[9]</sup> PJS presenting with intestinal obstruction due to jejunoileal intussusception in a 38-year-old male from India,<sup>[10]</sup> obstructing hamartomatous polyps in duodenum and small bowel in a 53-year-old male from US,<sup>[11]</sup> gastrointestinal cancers developing in first degree relatives in a PJS family<sup>[12]</sup> and few others.<sup>[13,14]</sup> All these reports bear consistency in presenting features of intestinal obstruction followed by polypectomies/gut resections, which revealed architectural distortions suggestive of PJS on histopathological examination. Although positive family history or mucocutaneous pigmentations are documented criteria for a PJS diagnosis,<sup>[1]</sup> they are rather infrequently observed in literature review.

## CONCLUSION

The low incidence of this disorder should not be an excuse for reluctance to perform screening in at risk

individuals, especially first degree relatives of the patient. Life-long screening for malignancy can sufficiently reduce morbidity and mortality in these patients. Finally, whether the association of maltoma with PJS increases the risk for malignancy needs to be documented and screening procedures adapted accordingly.

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