Bangladesh Crit Care J March 2025; 13 (1): 56-59

Case Report

Peutz-Jeghers Syndrome: A Rare Case Report

Mohammad Shamsul Arfin[1]*, Khan Md. Nazmus Saqeb[2]*, Mohammad Baharul Alam[3], Rebeka Razzaque[4]

DOI: https://doi.org/10.3329/bccj.v13i1.81307

Abstract:

Peutz-Jeghers syndrome (PJS) is a hereditary polyposis syndrome of the gastro intestinal tract with multiple hamartomatous polyps affecting almost any part of the gastrointestinal tract with pigmentation around lips and buccal mucosa. It is inherited in an autosomal dominant pattern. Most patients present with complications induced by the polyps like ulceration, anemia, bowel intussusception and sometimes malignant transformation of the polyps. Moreover, these patients have an increased risk of malignancy in other non-gastrointestinal sites as well. Here we present a case of a 29 years old male who presented with recurrent bouts of melaena. The patient had perioral pigmentations. Endoscopy upper GI and colonoscopy revealed multiple polyps all over the GIT. Histologically they were confirmed to be hamartomas, suggesting a diagnosis of PJS. Aggressive screening is recommended in patients with Peutz-Jeghers syndrome to prevent malignancy. Upper GI endoscopy and colonoscopy along with ileoscopy should be performed every two years from 10 years of age. Surveillance for cancers outside GIT including testicular and breast examinations once in a year should be introduced in the second decade of life.

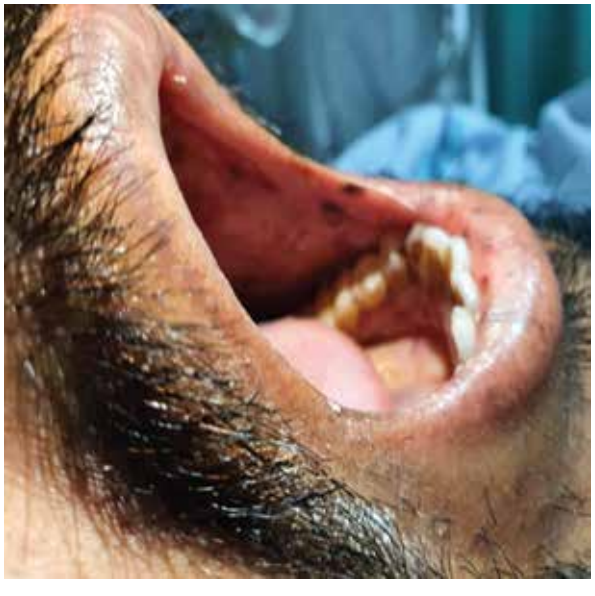
Keywords: *Peutz–Jeghers syndrome (PJS), Familial polyposis syndromes.*

Introduction

Peutz-Jeghers syndrome (PJS) was first reported by Peutz in 1921[1,2], and subsequently, detailed case PJS by Jeghers, McKusick, and Katz in 1949[3]. It[4,5] is a rare disorder characterized by typical pigmented perioral macules, pigmented spots in the buccal mucosa which are present in 90% of patients, and multiple hamartomatous polyps predominantly in the gastrointestinal (GI) tract[6]. Hamartomas can range from 5 to 50 mm in diameter (median size, 35 mm). Hamartomas are associated with intussusception, bleeding, anemia, and obstruction[7].In addition to polyposis, the risk of gastrointestinal and extra-gastrointestinal malignancies is significantly increased in PJS patients[8]. The relative risk of dying from a gastrointestinal cancer is 13 times greater. The risk of any other malignancy (especially cancer of the reproductive organs and breast, and also of the pancreas and lung) is 9 times greater than in the general population[9]. The gene was localized to chromosome 19p34p36 and is known as STK 11, a serine-threonine kinase involved in growth control regulation. On the other hand, not all patients with PJS have a mutation in this gene[10], suggesting other mutations.

Case history:

A young man, referred from Sylhet came to our hospital with the complaints of recurrent bouts of melaena for a month. Physical examination revealed severe anemia along with mucocutaneous pigmentations over the lips and buccal mucosa (Figure 1), no other lesions could be identified in the anal region or on the hands and feet. The abdominal examination was unremarkable with no organomegaly, mass or ascites.



1 Professor, Senior Consultant & Head, Dept. of Gastrointestinal, Hepatobiliary & Pancreatic diseases, Bangladesh Specialized Hospital, Shyamoli, Dhaka, Bangladesh.

2. Associate Professor & Head, Dept. of Gastrointestinal, Hepatobiliary & Pancreatic diseases, Jahurul Islam Medical College & Hospital, Bajitpur, Kishoreganj, Bangladesh.

3. Associate Consultant, Dept. of Gastrointestinal, Hepatobiliary & Pancreatic diseases, Bangladesh Specialized Hospital, Shyamoli, Dhaka, Bangladesh.

4. Specialist, Dept. of Gastrointestinal, Hepatobiliary & Pancreatic diseases, Bangladesh Specialized Hospital, Shyamoli, Dhaka, Bangladesh.

*Both Professor Dr. Mohammad Shamsul Arfin and Dr. Khan Md. Nazmus Saqeb had equal contributions to the case report & both will be considered as first authors.

Corresponding Author:
Dr. Khan Md. Nazmus Saqeb
Associate Professor
Dept. of Gastrointestinal, Hepatobiliary & Pancreatic diseases
Jahurul Islam Medical College & Hospital
Bajitpur, Kishoreganj, Bangladesh.
Mail: drsaqebk59@gmail.com
ORCID ID: https://orcid.org/0000-0002-8080-947X

Received : February 02, 2025; Accepted : March 01, 2025

Figure 1: Typical mucocutaneous pigmentation around lips of our patient.

Laboratory data confirmed anemia with no other blood, urine or stool changes. We proceeded with an upper gastrointestinal endoscopy, which revealed large number of variable sized polyps in the stomach. Large number of variable sized polyps were also seen in the bulb, post bulbar area & proximal jejunum. (Figure 2 & 3). Additionally, colonoscopy was performed and large number of polyps in clusters were seen throughout the colon and rectum along with multiple small polypoid lesions at the terminal ileum. Endoscopic polypectomy was done from stomach and duodenum. Colonoscopic polypectomy was done from colon and rectum. All the polyps were sent for histopathology.

A CT scan was done to check if there was any other extraintestinal site involvement or complications. Tumor markers were checked: CA19.9 and CEA were normal. Histopathology of the resected polyps revealed arborization of smooth muscle bundles in muscularis mucosae covered with normal villi and was defined as a hamartoma, thus confirming the clinical diagnosis of PJS.

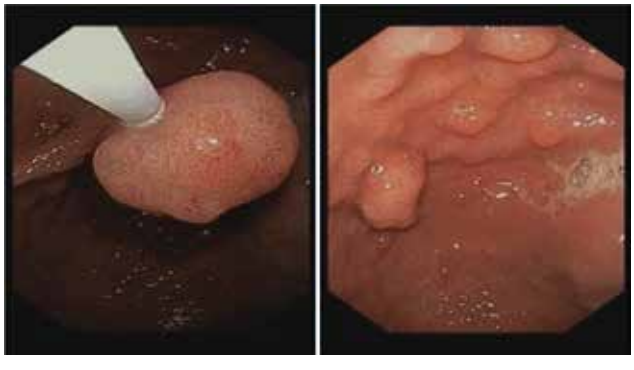


Figure 2: Multiple polyps at the stomach at upper GI endoscopy and endoscopic polypectomy.

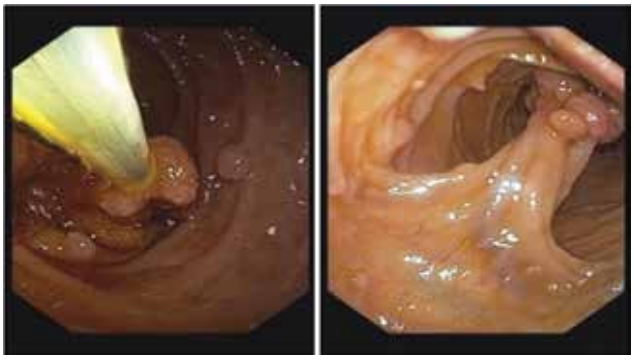


Figure 3: Multiple polyps at the duodenum at upper GI endoscopy and endoscopic polypectomy.

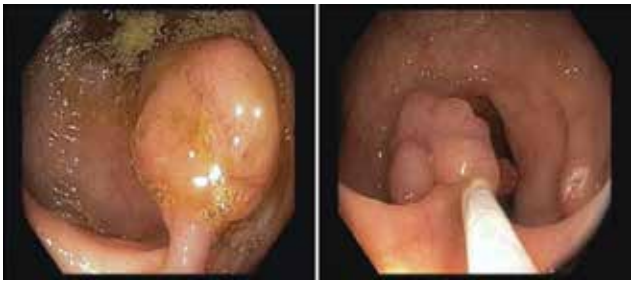


Figure 4: Multiple polyps at the rectum at colonoscopy and colonoscopic polypectomy.

A further detailed family history was obtained. Patients father died of colorectal carcinoma (could not be confirmed if it was Peutz- Jeghers associated). His elder brother died of rectal carcinoma at the age of 44 and his younger brother is currently undergoing endoscopic polypectomy procedure in Sylhet. His younger brother has lip pigmentations as per the patient's statement. Her mother first noted pigmented spots around his lips when he was 9 years of age. Genetic study was not performed. The patient was placed on a multi-session polypectomy schedule and was placed on a regular cancer surveillance program for cancers outside the GIT.

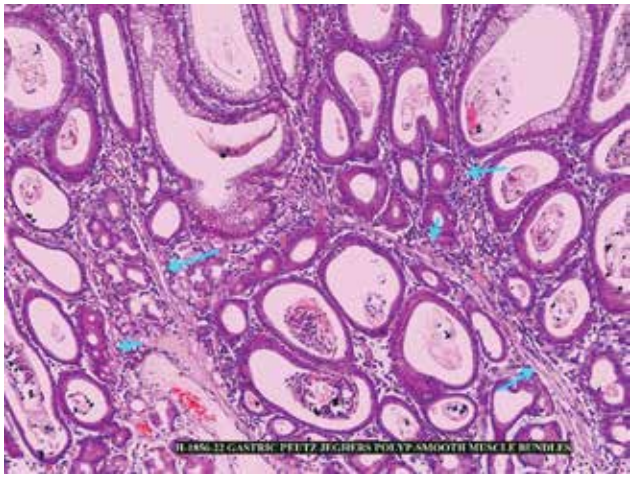


Figure 5: The histopathological findings reveal hamartoma, with a typical gastric PJS polyp demonstrating an arborizing pattern of growth of smooth muscle (HE staining, magnification 4×).

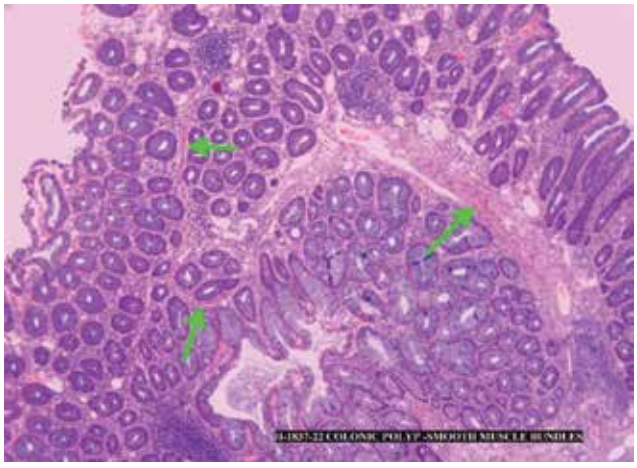


Figure 6: The histopathological findings reveal hamartoma, with a typical colonic PJS polyp demonstrating an arborizing pattern of growth of smooth muscle (HE staining, magnification 4×).

Discussion

Peutz–Jeghers syndrome is an autosomal dominant syndrome characterized by gastrointestinal hamartomatous polyposis associated with oral and anal mucocutaneous pigmentations.[11] The syndrome also manifests extraintestinal hamartomatous polyps[12]. Jeghers polyps occur most numerously in the small intestine but frequently in the colon and stomach, only a few cases have been reported in the duodenum.[12] The World Health Organization (WHO) criteria for diagnosing this rare disorder are[13]:

1. Three or more polyps, which show histological features consistent with PJS.
2. A family history of PJS with any number of PJPs.
3. A family history of PJS with characteristic mucocutaneous pigmentation.
4. Characteristic mucocutaneous pigmentation with any number of PJPs.

Patients with PSJ carry a very high risk of gastrointestinal adenocarcinoma along with exaggerated extraintestinal carcinoma risk- namely pancreatic, breast, testis, and ovary. Compared with the normal population, PJS subjects have a relative risk of 15 for developing any such type of malignancy.[9] We report a case of Peutz–Jeghers syndrome in a patient with intermittent melaena and describes the clinical characteristics of PJS.

The actual etiology of PSJ is unclear. Most commonly mutated gene that has been identified is STK11. STK 11 gene is a tumor suppressor gene which codes a serine/ threonine kinase.[14] Mutation of this gene leads to loss of tumor suppression attribute of the gene and hence development of the polyps, associated in many cases with cancer development. So, in comparison to the general population, individuals have a higher incidence rate of malignant tumors. Colorectal cancer is the most commonly associated carcinoma with PSJ, followed by breast, gastric, small bowel, and pancreatic carcinoma.[15]

Patients should be instructed on the need for cancer surveillance- follow-up abdominal ultrasound examination (liver gallbladder, pancreas, spleen) every six months and upper gastrointestinal endoscopy every three years according to the guidelines for PJS. The differential diagnosis of SPJ arises mainly with other hereditary family polyposis as familial adenomatous polyposis, Gardner's syndrome and generalized juvenile polyposis[16].

Many new therapies are now going through trial on animals to estimate its effectiveness in reducing polyp growth. PSJ polyps overexpress COX2 enzyme, hence, celecoxib (a potent COX 2 inhibitor) showed halt of polyp development in mice models successfully. However, trail on humans has yet awaiting.[17] A combined surgical and endoscopic procedure allows effective clearance of the multiple hamartomatous polyps from the entire length of the small bowel reported and considered superior in terms of quality of life. Surgical intervention is necessary when the condition is complicated by its known complications e.g. Gastrointestinal bleeding, obstruction, or intussusception or the polyp count and size is too big to be removed by endoscopy.

Most publications recommend polypectomy for polyps in the stomach or colon that are greater than 1 cm in size noted during endoscopic surveillance. Surgery has been recommended for symptomatic or rapidly growing polyps or asymptomatic polyps greater than 1–1.5 cm in size. Some experts suggest a clean sweep. This can be facilitated by concomitant interoperative endoscopy with polypectomy or, in the case of larger polyps, enterotomy. The clean sweep approach appears to decrease the need for recurrent small bowel surgery. Recently, the use of double balloon enteroscopy for removal of small bowel PJS polyps has been reported and might decrease the need for laparotomy[18]. Lifelong cancer surveillance is recommended. At the age of 8–10 years, surveillance for stomach and small bowel polyposis should start, and it should continue every 2–3 years[19].

Conclusion

Peutz Jeghers patients are rare clinical entity, when diagnosed, should undergo periodic surveillance. Removal of larger polyps aims to reduce the likelihood of complications in Peutz-Jeghers. Hence, surveillance for gastric and small-bowel polyposis should begin at age 8-10 years and continue at 2-3–year intervals[20]. Other family members should be screened as well. Early diagnosis, endoscopic or surgical resection and regular follow-up can have a significant positive effect on its prognosis, delaying/ preventing the complications associated with PSJ as well as transformation of the polyps into overt malignancy.

References

1. Tweedie JH, McCann BG. Peutz-Jeghers syndrome and metastasizing colonic adenocarcinoma. Gut 1984; 25: 1118–1123.
2. Chowdhry S, Umrigar DD, Yadav N. Peutz-Jeghers syndrome in a child presenting with acute abdomen: a case report, Asian J. Dermatol 2015; 8: 20–24.

3. Foley TR, McGarrity TJ, Abt AB. Peutz-Jeghers syndrome: a clinicopathologic survey of the “Harrisburg family” with a 49-year follow-up, Gastroenterology 1988; 95: 1535–1540.

4. Peutz JL. On a very remarkable case of familial polyposis of the mucous membrane of the intestinal tract and nasopharynx accompanied by peculiar pigmentation of the skin and mucous membrane. Ned Tijdschr Geneeskd 1921; 10: 134-46.

5. Jeghers H, McCusick VA, Katz KH. Generalized intestinal polyposis and melanin spots of the oral mucosa, lips and digits. A syndrome of diagnostic significance. N Engl J Med 1949; 241: 993-1005.

6. Tomlinson IP, Houlston RS. Peutz-Jeghers syndrome. J Med Genet 1997; 34 (12): 1007-11.

7. Latchford A, Cohen S, Auth M, Scaillon M, Viala J, Daniels R, et al. Management of Peutz-Jeghers syndrome in children and adolescents, J Pediatr Gastroenterol Nutr. 2019; 68: 442–452.

8. Boardman LA, Thibodeau SN, Schaid DJ, Lindor NM, McDonnell SK, Burgart LJ, et al. Increased risk for cancer in patients with the Peutz-Jeghers syndrome. Ann Intern Med 1998; 128 (11): 896-9.

9. Spigelman AD, Murday V, Phillips RK. Cancer and the Peutz-Jeghers syndrome. Gut 1989; 30 (11): 1588-90.

10. Boardman LA, Couch FJ, Burgart LJ, Schwartz D, Berry R, McDonnell SK, et al. Genetic heterogeneity in Peutz-Jeghers syndrome. Hum Mutant 2000; 16: 23-30.

11. Arber N, Moshkowitz M. Small bowel polyposis syndromes. Curr Gastroenterol Rep 2011; 13:435–41.

12. Giardiello FM, Trimbath JD. Peutz–Jeghers syndrome and management recommendations. Clin Gastroenterol Hepatol 2006; 4:408–15.

13. Schreibman IR, Baker M, Amos C, McGarrity TJ: The hamartomatous polyposis syndromes: a clinical and molecular review. Am J Gastroenterol 2005, 100(2):476-490.

14. Hemminki A, Markie D, Tomlinson I, et al. A serine/threonine kinase gene defective in Peutz–Jeghers syndrome. Nature 1998; 391:184–7.

15. van Lier MG, Wagner A, Mathus-Vliegen EM, et al. High cancer risk in Peutz–Jeghers syndrome: a systematic review and surveillance recommendations. Am J Gastroenterol 2010; 105:1258–64.

16. Nakagawa H, Koyama K, Tanaka T, Miyoshi ., Ando H, Baba S. et al, 1998, Localization of the gene responsible for PeutzJeghers syndrome within a 6-cM region of chromosome 19p13.3. Hum Genet , 102 ,pp. 203-206.

17. https://rarediseases.org/rare-diseases/peutz-jeghers-syndrome/

18. Chen HY, Jin XW, Li BR, Zhu M, Li J, Mao JP, et al. Cancer risk in patients with Peutz-Jeghers syndrome: a retrospective cohort study of 336 cases, Tumour Biol. 2017 June; 39(6): 1010428317705131. doi: 10.1177/1010428317705131.

19. Wang H, Luo T, Liu WQ, Huang Y, Wu XT, Wang XJ. Clinical presentations and surgical approach of acute intussusception caused by Peutz–Jeghers syndrome in adults. J Gastrointest Surg. 2011;15(12):2218–25.

20. Tan VK, Koh PK, Loi CT, Eu KW, Tang CL. Peutz-Jeghers syndrome: data from the Singapore Polyposis Registry and a shifting paradigm in management. Ann Acad Med Singapore. 2010 Jan. 39 (1):17-21.