DOI: NJLM/2015/13284:2064

Case Report

Pathology Section

Solitary Peutz-Jeghers Polyp of Jejunum with Dysplasia Presenting as Intussusception

DEEPTI AGARWAL, SWARN KAUR SALUJA, DHEERAJ PARIHAR, PARVEEN KUNDU

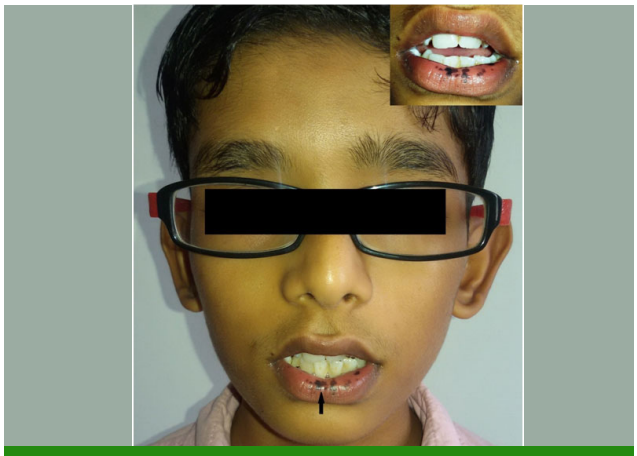
ABSTRACT

Peutz-Jeghers syndrome is inherited as an autosomal dominant disorder presenting as hamartomatous polyps in small bowel with mucocutaneous pigmentation. We here present a case of 14 years old male presenting with symptoms of intestinal obstruction and mucocutaneous pigmentation along with family history of similar rashes, in both his mother and brother.

Keywords: Hamartomatous polyps, Jejunal polyps, Mucocutaneous pigmentation

CASE REPORT

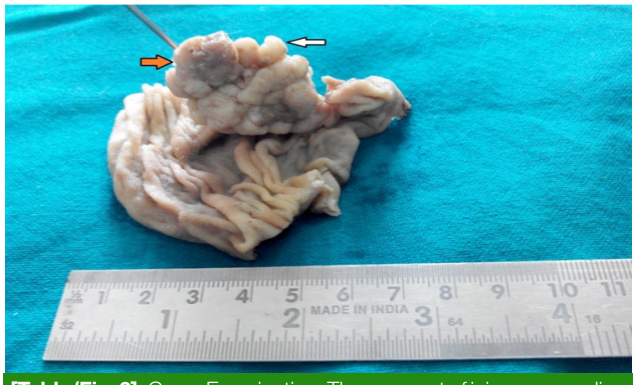
A 14 years old male presented with symptoms of abdominal pain and loss of appetite in Surgical outpatient Department since two days. He had a history of similar episodes of abdominal pain off and on since past 3 months. Ultrasonography revealed features of intestinal obstruction due to small bowel mass. CT scan of the abdomen revealed intussusception of jejunal loop over a single polypoidal mass. The patient had a mucocutaneous pigmentation over the lower lip [Table/Fig-1]. Both his mother and brother had a history of intestinal polyps with mucocutaneous pigmentation. A diagnosis of recurrent small bowel obstruction was made and a laparotomy was performed to relieve the obstruction. The affected bowel segment was resected with anastamosis of the normal bowel and the specimen was sent for histopathological evaluation. The patient was discharged in good health with no recurrence of polyps on sonographic examinations at 6 month and 1 year follow up.



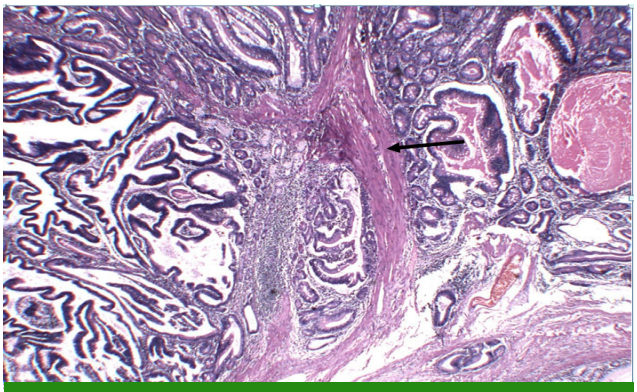
[Table/Fig-1]: Mucocutaneous rashes on lower lip (arrow). A Bluish hue is observed in the pigmented lesion (Inset)

Gross Examination: A segment of jejunum measuring 7cm in length was received for histopathological evaluation. A pedunculated polypoidal growth was seen in the jejunum measuring 5x5x4 cm [Table/Fig-2]. The outer surface showed broad papillae with cerebriform appearance.

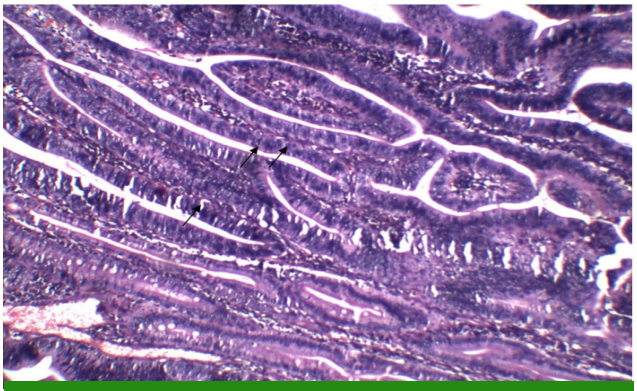
Microscopic examination: The polyp had a central core of smooth muscle that showed tree-like branching and becoming progressively thinner as they reach the polyp surface polyp i.e. arborizing muscular framework [Table/Fig-3]. It was covered by the mucosa native to the region, heaped into folds producing a villous pattern. At places the lining epithelium exhibited dysplastic changes [Table/Fig-4].



[Table/Fig-2]: Gross Examination: The segment of jejunum revealing a pedunculated polypoidal growth is seen 5x5x4 cm (Orange arrow). Outer surface shows broad papillae with cerebriform appearance (White arrow)



[Table/Fig-3]: Microscopic examination (H&E) 20X: Smooth muscle shows tree-like branching or arborizing muscular framework (arrow)



[Table/Fig-4]: Microscopic examination (H&E) 40X: Lining epithelium exhibiting dysplastic changes (arrows)

A final diagnosis of hamartomatous polyp with dysplastic changes was reached after correlation with the clinical and histopathological findings, thus establishing the diagnosis of Peutz-Jeghers syndrome with dysplastic changes in polyp.

DISCUSSION

Peutz-Jeghers Syndrome (PJS) is a rare, familial, autosomal dominant disorder that presents with multiple gastrointestinal hamartomatous polyps and mucocutaneous hyperpigmentation due to mutation in gene encoding Serine Threonine Kinase (STK 11) also known as LKB1 [1]. The incidence in general population varies from 1 in 8000 to 1 in 30,000 live births according to the literature [2]. The common clinical presentation includes intussusception, bleeding or bowel obstruction due to the polyploidal mass in small intestine. Despite their benign nature, there is a certain increased risk of progression to malignancy in some cases, observed in follow-up of the patients in few reports [3]. In recent studies an increased risk of gastrointestinal and extra- gastrointestinal cancers (breast and gynecological cancers in females) has been observed. In present case, apart from the classical clinical presentation, various other atypical presentations and their criteria have been discussed.

The clinically defined criteria to diagnose the varied presentations in Peutz-Jeghers polyposis includes: (i) Three or more histologically confirmed Peutz-Jeghers polyps, or (ii) Any number of Peutz-Jeghers polyps with a family history of PJS, or (iii) Characteristic prominent mucocutaneous pigmentation with a family history of PJS, or (iv) Any number of Peutz-Jeghers polyps and characteristic prominent mucocutaneous pigmentation [4]. Commonly, the polyps of PJS are hamartomatous and involve jejunum, ileum, colon, stomach, duodenum and appendix in decreasing order of frequency. These polyps are derived from glandular epithelium with branching smooth muscles from muscularis mucosa. They are usually multiple in number, can be sessile or pedunculated and usually occur before the age of 20 years in 50% of the cases. Sometimes, the polyps reach up to 100 in number, though our case had a solitary jejunal polyp with dysplasia. The size of polyp usually range from few mm to over 7 cm; most measuring within 0.5 to 3.0 cm in diameter [4, 5].

The pigmentation in PZ polyposis is characteristic with freckle like pigmented macules found on lips, around mouth, mucosa of nose, conjunctiva or rectum, skin of finger and toes in more than 90% cases. It is secondary to pigment laden macrophages in the dermis. The diagnosis is more likely possible in pediatric patients as pigmented lesions may fade at puberty [6]. In a case series reported by Suda et al ., solitary Peutz-Jeghers-type hamartomatous polyps presented without pigmented spots in 87% cases, involved only one organ in the gastrointestinal tract, and showed a predilection for males in 74% cases. They also observed that solitary hamartomatous polyps are histologically similar to those in Peutz-Jeghers syndrome [7]. It has been described in literature that solitary polyps exhibit less branching of the muscularis mucosae than in the familial form (with multiple polyposis), as was evident in our case also . If any area of dysplasia is located in the submucosa and muscularis mucosa, a mistaken diagnosis of cancer may be concluded during histopathology evaluation [4]. In our case, the features of dysplastic changes were present in the lining epithelium.

Differential diagnosis of PJ polyp includes pseudoinvasion, adenomas, juvenile and inflammatory polyps. Epithelial misplacement i.e. pseudoinvasion extending upto serosa may be over diagnosed as a well differentiated adenocarcinoma. Features like lack of cellular atypia, presence of mucinous cysts and haemosiderin deposition go against the diagnosis of a neoplastic process. Juvenile polyps lack band of smooth muscles which is seen in adenomas and PJ polyp. Presence of a well-demarcated focus of dysplasia favors adenoma while haphazard arrangement of glands is a feature seen in inflammatory polyps [4].

The risk of developing a certain malignancy in PJ polyposis increases to 93% by the age of 64 years. The overall risk of death by cancer in PJS patients is about 50% at 58 years of age [4, 5]. The risk of gastrointestinal cancer is 15 times greater for patients with PJS than for general population. However, due to the rarity of the syndrome, there is little documentation on the prognosis. Open or endoscopic polypectomy is a

recommended conservative procedure for pedunculated polyps, as repeated resections of bowel segments lead to short bowel syndrome in long course [8,9]. Furthermore, regular screening is recommended in suspected cases with familial history to limit the morbidity and severity of disease.

CONCLUSION

Patients with PJS are prone to develop digestive or extra digestive cancers especially when the age of onset of symptoms is early. Presence of more than 4 polyps, size more than 1 cm and the presence of dysplastic lesions in polyps increases the risk of malignancy. Surgical excision is the treatment of choice in presence of multiple polyps and complications like hemorrhage or obstruction.

REFERENCES

[1] Hearle N., Schumacher V., Menko F.H. et al. STK11 status and intussusception risk in Peutz-Jeghers syndrome. *J. Med. Genet.* 2006; 43(8): e41.

[2] Choi S H, Park Y J, Park J G. Peutz-jeghers syndrome- a new understanding. *J Korean Med* Sc.1999; 14(1):2-7.

[3] Van Lier MGF, Wagner A, Mathus-Vliegen EMH, Kuipers EJ, Steyerberg EW, Van Leerdam ME. High cancer risk in Peutz-Jeghers syndrome: a systematic review and surveillance recommendations. *American Journal of Gastroenterology.* 2010; 105 (6):1258–64.

[4] Choudhury T, Enam S, Begum F, Taufiq M, Kamal M. Peutz-Jeghers polyp: a retrospective study on twelve cases received at the department of Pathology , Bangabandhu Sheikh Mujib Medical University. *BSMMU J.* 2012; 5(1):12-17

[5] Giardiello FM, Brensinger JD, Tersmette AC, et al. Veryhigh risk of cancer in familial Peutz-Jeghers syndrome. *Gastroenterology.* 2000;119(6):1447–53.

[6] Beggs AD, Latchford AR, Vasen HFA, et al. Peutz-Jeghers syndrome: a systematic review and recommendations for management. *Gut.* 2010; 59(7):975–86.

[7] Nakayama H, Fujii M, Kimura A, Kajihara H. A Solitary Peutz-Jeghers-type hamartomatous polyp of the rectum: report of a case and review of the literature. *Jpn J Clin Oncol* 1996; 26(4):273-76.

[8] Gama-Rodrigues JJ. Intestinal intussusception and occlusion caused by small bowel polyps in the Peutz-Jeghers Syndrome. management by combined intraoperative enteroscopy and resection through minimal enterostomy: case report. *Re. Hosp. Clin. Fac. Mae. S. Paulo.* 2000; 55(6):219–24.

[9] Cunningham JD, Vine AJ, Karch L, Aisenberg J. The role of laparascopy in the management of intussuseption in Peuty-Jeghers syndrome: case report and review of the literature. *Surgical Laparoscopic Endoscopy.* 1998; 8(1):17–20.

AUTHOR(S):

1. Dr. Deepti Agarwal
2. Dr. Swarn Kaur Saluja
3. Dr. Dheeraj Parihar
4. Dr Parveen Kundu

PARTICULARS OF CONTRIBUTORS:

1. Associate Professor, Department of Pathology, BPS Government Medical College for Women, Khanpur Kalan, Sonipat, Haryana, India.
2. Professor & Head, Department of Pathology, BPS Government Medical College for Women, Khanpur Kalan, Sonipat, Haryana, India.
3. Assistant Professor, Department of Pediatric Surgery, BPS Government Medical College for Women, Khanpur Kalan, Sonipat, Haryana, India.
4. Assistant Professor, Department of Pathology, BPS Government Medical College for Women, Khanpur Kalan, Sonipat, Haryana, India.

NAME, ADDRESS, E-MAIL ID OF THE CORRESPONDING AUTHOR:

Dr. Deepti Agarwal,
Associate Professor, Department of Pathology,
BPS Government Medical College for Women,
Khanpur Kalan, Sonipat, Haryana, India.
E-mail: drdeepti75@gmail.com

FINANCIAL OR OTHER COMPETING INTERESTS: None.

Date of Publishing: **Oct 01, 2015**