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Case Report

Peutz-Jeghers Syndrome Presenting with Intussusception in a Young Woman: A Case Report

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ABSTRACT

The Peutz-Jeghers syndrome (PJS) is an autosomal dominant disorder characterized by hamartomatous polyposis of the gastrointestinal tract, melanin pigmentation of the skin and mucous membranes and an increased risk for cancer. The polyps can cause anemia and intestinal obstruction and rarely intussusception. We present a case of young female with a history of pain in abdomen and anemia. Patient had mucocutaneous pigmentations on the lips and buccal mucosa. USG was suggestive of inflammatory bowel lesion of size 9.9x6.8x4 cm. CT scan revealed target sign and ribbon sign in right side of abdomen suggestive of intussusception. Intraoperatively, jejunal polyps were found to be the cause of jejuno-jejunal intussusception. Histopathology revealed hamartomatous polyps of Peutz-Jeghers syndrome.
Our interest in this case is due to the uncommon presentation of PJS as intussusception in adult and the observation of three cases in the same family, all three of them had Peutz-Jeghers polyp, while their mother had perioral pigmentation only. It is suggested that any patient presenting with ileus attacks and anemia should be investigated for polyps and mucocutaneous pigmentations of the PJS.

Keywords: Peutz-Jeghers syndrome (PJS), Intussusception.

INTRODUCTION

Peutz-Jeghers syndrome (PJS) is a rare autosomal dominant disorder characterized by hamartomatous polyps throughout the gastrointestinal tract and characteristic mucocutaneous pigmentation. Its incidence has been estimated as one in 120,000 births. It affects males and females equally. [1] The relative risk of dying from a gastrointestinal cancer is 13 times greater and the risk of any other malignancy (especially cancer of the reproductive organs and breast, and also of the pancreas and lung) is nine times greater in patients of PJS than in the general population. [2]

Most of the polyps of PJS are confined to the small bowel (60% to 90%) and the colon (50% to 64%). These polyps may also be found at extra-intestinal sites. [3] They have been reported to cause gastrointestinal bleeding in 14% of patients and recurrent intestinal obstructions in 43% of patients. However, adult intussusception is relatively rare; only 5% to 16% of cases are in adults, and they contribute to only 1% of all causes of intestinal obstruction. The most seen types of intussusception are jejunojejunal, jejunoileal, ileoileal and ileocolic. [4]

We present here an uncommon presentation of Peutz-Jeghers syndrome in

a young female with a very strong family history.

CASE REPORT

A 26-year-old woman was presented with acute abdominal pain and vomiting. This colicky pain was located in right side of abdomen and was non-radiating in nature. On general examination, patient had pallor. Mucocutaneous pigmentation was noted on her lips and oral mucosa (Figure 1). Local examination revealed tenderness in right lower abdomen. An approximately 8cm-diameter mass was palpable and bowel sounds were significantly increased at this location. Routine laboratory investigations were normal except for anemia. Patient underwent ultrasonography (USG), followed by CT scan. USG showed hypoechoic, well demarcated lesion of size 9.9x6.8x4 cm in right iliac fossa suggestive of inflammatory bowel lesion. CT scan revealed bowel within bowel showing target sign and ribbon sign suggestive of intussusception. The CT did not reveal any other polyps in her gastrointestinal tract. Patient had a strong family history of Peutz-Jeghers syndrome. Her elder sister and brother were operated in past for jejunal polyps, histology of which showed hamartomatous polyps of PJS. Also, her mother had mucocutaneous pigmentations on oral mucosa and feet, but did not have any polyp.

A laparotomy was done for definitive management of intussusception. On exploration, a jejunojejunal intussusception was found 50cms from duodenojejunal junction. Intussusception was reduced and found to be because of multiple jejunal polyps. Resection of the involved jejunal segment was done and the cut ends were anastomosed. Postoperative period was uneventful.

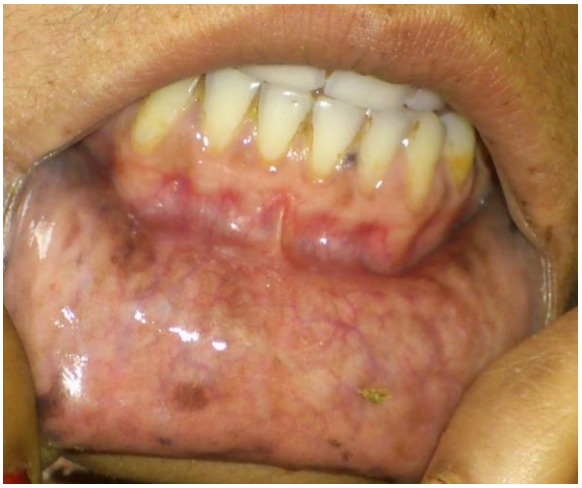

Figure-1: Dark-brown pigmented maculae on lower lip

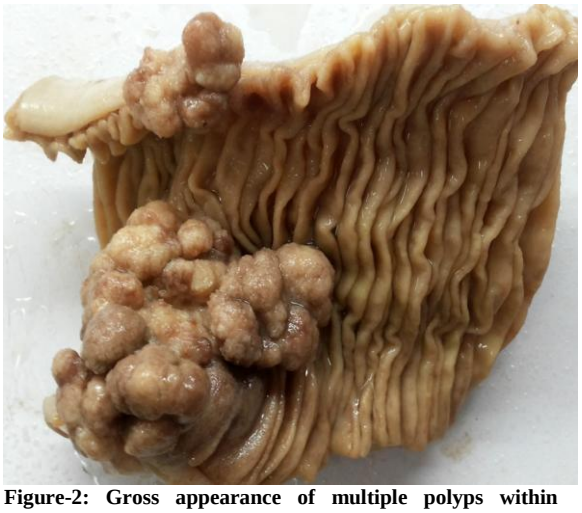

Figure-2: Gross appearance of multiple polyps within intussusceptum of jejunum

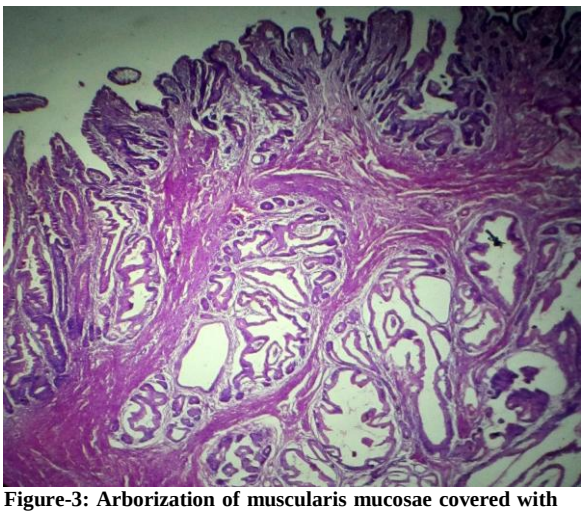

Figure-3: Arborization of muscularis mucosae covered with normal villi (H&E, 10x)

Gross pathologic findings of the specimen showed 15cm long segment of jejunum with multiple polyps within it. Larger one measuring 7x6x2 cm and smaller measuring 2.8x2x1cm (Figure 2).

Histopathological examination revealed a hamartomatous polyp which has convoluted, elongated glands and an arborizing pattern of growth and consist of a branching framework of smooth muscle and connective tissue lined by normal epithelium, without evidence of any malignancy (Figure 3&4).

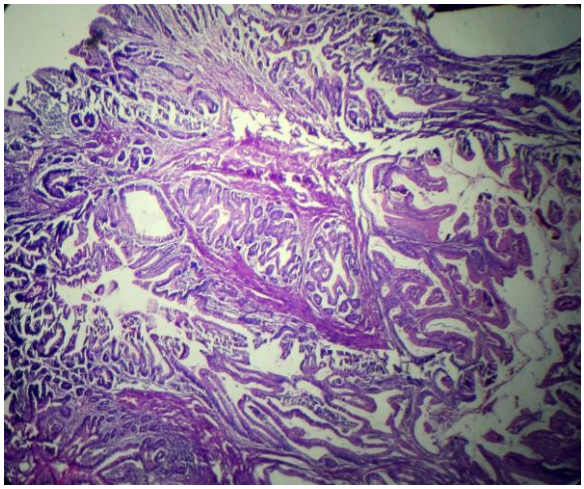

Figure-4: Hamartomatous polyp with presence of smooth muscular fibers dissociated and branched between glandular structures (H&E, 10X)

DISCUSSION

PJS is a complex hereditary condition characterized by hamartomatous polyps of the gastrointestinal tract and mucocutaneous pigmentation. It is caused by a germline mutation in the serine/threonine kinase 11 gene. A clinical diagnosis of PJS may be made when any one of the following conditions is present in a single individual: two or more histologically confirmed PJ polyps; any number of PJ polyps detected in one individual who has a family history of PJS in a close relative; characteristic mucocutaneous pigmentation in an individual who has a family history of PJS in a close relative; any number of PJ polyps in an individual who also has mucocutaneous pigmentation. Fewer than 5% of patients with PJS lack the abnormal mucocutaneous pigmentation, and fewer than 5% of patients with the pigmentation have no PJ polyps. [3]

Cutaneous markers of systemic disease are vital for clinicians to recognize. Differential diagnosis of familial lentiginosis syndromes includes Peutz-Jeghers syndrome, Carney Complex, the PTEN hamartomatous syndromes, and LEOPARD/Noonan syndrome. [5]

The pigmentation of PJS may be present at birth but usually develop in early childhood. They are most commonly found around the mouth, nose, lower lip, buccal mucosa, hands and feet. Oral pigmentation is usually permanent, but the maculae on the lips and skin may fade after puberty. [6]

Peutz-Jeghers polyps are commonly found in the small bowel, colon, and stomach in decreasing frequency. The pedunculated nature and the large size of the polyps can lead to intussusception in the small bowel. [7] Intussusception is uncommon in adult patients with PJS. The majority of PJS intussusceptions reported in the literature are ileal or jejunal. Colo-colonic intussusception is reported in only a few cases. [3] The diagnosis of intussusception caused by PJS should be based on a history of PJS and physical examination. Abdominal distention and local tenderness are the most frequent findings. We located a palpable mass in our patient but an abdominal mass has been noted in only 12.5% of cases. [8]

As a readily available and inexpensive method, ultrasonography may demonstrate small bowel polyps in patients with PJS and typically show a “doughnut” sign suggestive of an intussusception. CT scan may be the most accurate modality for diagnosis of adult intussusceptions. [8] CT may help to delineate the precise location of polyps and differentiate between lead point and non-lead point intussusception. Endoscopy has a distinct role in the diagnosis and treatment of intussusception. Endoscopic polypectomy and double-balloon enteroscopy can avoid short bowel syndrome in the case of multiple intussusception. However, when this is not possible, laparoscopy can be a

safe and effective therapeutic alternative. [3] Any polyp that is larger than 1.5cm should be removed if possible, as it generally causes intussusceptions. [1] Therefore, we performed a laparotomy to reduce the intussusception and resect her bowel.

CONCLUSION

Diagnosis of intussusception in adults is usually delayed because of the patient's nonspecific and chronic symptoms. A thorough review of the patient's history, physical examination, and radiological and endoscopic findings are critical in the case of intussusceptions in PJS, which is an uncommon presentation. Early diagnosis is important as affected patients and at-risk family members should be offered surveillance from an early age for early detection of GI tract malignancies.

REFERENCES

1. Shrivastava A, Gupta A, Gupta A, and Shrivastava J. Unusual Presentation of Intussusception of the Small Bowel with Peutz-jeghers Syndrome: Report of a Case. J Clin Diagn Res. 2013 Oct; 7(10): 2296–2297.
2. Homan M, Dolenc Strazar Z and Orel R. Peutz-Jeghers syndrome. A case report. Acta Dermatovenerol Alp Pannonica Adriat. 2005 Mar; 14(1):26-9.
3. Ozer A, Sarkut P, Ozturk E, and Yilmazlar T. Jejunoduodenal intussusception caused by a solitary polyp in a woman with Peutz-Jeghers syndrome: a case report. J Med Case Rep. 2014; 8: 13.
4. Thakker HH, Joshi A, Deshpande A. Peutz-Jegher's syndrome presenting as jejunoileal intussusception in an adult male: a case report. Cases J. 2009; 2:8865–8868.
5. Maya B. Lodish and Constantine A. Stratakis. The differential diagnosis of familial lentiginosis syndromes. Fam Cancer. 2011 Sept; 10(3):481-490.
6. Georgescu EF, Ligia Stănescu, Cristiana Simionescu, Iuliana Georgescu. Peutz–Jeghers syndrome: case report and literature review. Romanian Journal of Morphology and Embryology. 2008; 49(2):241–245.
7. McGarrity TJ, Kulin HE, Zaino RJ. Peutz–Jeghers syndrome. Am J Gastroenterol. 2000; 95:596–604.
8. Barussaud M, Regenet N, Briennon X, de Kerviler B, Pessaux P, Kohneh-Sharhi N, et al. Clinical spectrum and surgical approach of adult intussusceptions: a multicentric study. Int J Colorectal Dis. 2006; 21:834–39.

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