ISSN: 0975-3583, 0976-2833 VOL14, ISSUE 09, 2023

PEUTZ-JEGHERS SYNDROME: RARE CASE ENTITY WITH JEJUNAL POLYPS LEADING TO JEJUNOJEJUNAL INTUSSUSCEPTION

Authors: Dr Permeet Kaur Bagga, Dr Rimple Khatri, Dr Neetika Kaushal*,

Dr Rakesh Sharma

***Corresponding Author : Dr Neetika Kaushal**

INTRODUCTION

Peutz-Jeghers syndrome (PJS) is an autosomal dominant inherited polyposis syndrome characterized by multiple hamartomatous polyps in the gastrointestinal tract, associated with mucocutaneous pigmentation, especially around the mouth. It is a rare condition in most cases, attributed to mutation in STK11 gene [1]. Polyps vary in size (from few millimeters to approximately 7 cm) and location (mainly found in the small intestine, but also in extra-intestinal sites) and may occasionally be absent [2, 3]. In addition to polyposis, the risk of gastrointestinal and extra-gastrointestinal malignancies is significantly increased in PJS patients [4]. A regular lifetime surveillance is recommended after the polyp resection.

CASE REPORT

A young girl, 17 years of age was apparently well before she presented with diffuse abdominal pain, vomiting and fever. On examination the patient was toxic and restless with rapid pulse. Marked tenderness was present in the upper abdomen and a palpable, mobile firm to hard mass felt in periumblical area. Abdominal CECT revealed jejuno-jejunal intussusception. An emergency laparotomy was performed. Jejuno-jejunal intussusception showing approximately 30 cm of distal jejunum invaginating into the proximal jejunum was found. The intussusception could not be released, hence it was resected and sent for histopathology.

On gross examination**,** resected specimen of gut measured 62cm in length with dull appearing serosa and a perforation (Fig 1). On cutting 30 cm of intussuscepted gut loop the inner surface was homogenously grey brown. In the lumen of the gut, two pedunculated polyps were identified measuring 3x2x1.5cm and 4x1.7x1cm. (Fig 2)

Also received 3 resected polyps from proximal jejunum ranging from 1cm to 2.5cm in diameter which were green to reddish grey in color with irregular surface.

On histopathological examination, polyps showed characteristic complex architecture of intestinal type of glands supported by arborising network of connective tissue, thick bands of smooth muscle intermixed with lamina propria . (Fig.3)

Family history of Peutz-Jeghers Syndrome/any other polyposis syndrome was not there. On careful re-examination of the patient, multiple pigmented patches were seen over buccal mucosa (Fig. 4). She was discharged with a diagnosis of Peutz-Jeghers syndrome with an advice for regular follow ups.

ISSN: 0975-3583, 0976-2833 VOL14, ISSUE 09, 2023

On follow up colonoscopy, multiple rectal pedunculated polyps were seen and one of polyps was snared and sent for histopathology which showed classical histological features of Peutz-Jeghers syndrome.

DISCUSSION

PJS was initially documented by an English physician in 1895 [1] and has been recognized as a distinct clinical entity in 1949 by Jeghers who defined it as the coexistence of mucocutaneous pigmentation and gastrointestinal polyposis. The estimated incidence of the syndrome is between 1 in 50,000 to 1 in 2,00,000 live births [1]. It has been reported that one-third of patients are symptomatic by age of 10, and that half of them have experienced complications including intussusception, obstruction or bleeding, by age 20 [5]. In this case age of patient was 17 years and she presented with complication of intussusception. The gastrointestinal polyps which are hallmark of PJS are mostly found in small intestine but can also be seen in stomach and large intestine. In this case multiple polyps were presented in small intestine and on follow up colonoscopy, polyps were also seen in rectum.

Giardello et al [6] proposed diagnostic criteria for PJS which requires histopathological confirmation of hamartomatous gastrointestinal polyps and two of the following features: small bowel polyposis, positive family history and pigmented skin or mucosal brown macules. In this case hamartomatous gastrointestinal polyps and multiple pigmented patches were seen over buccal mucosa however family history was not positive.

PJS shows an autosomal dominant pattern of inheritance with both familial (80%) and sporadic (25%) transmissions. The most common gene involved was localized to chromosome 19p34-p36 and is known as STK11, a serine-threonine kinase involved in growth control regulation. Mutations of chromosomes 6q and 19q have been suggested in a few families [7,8]. The polyps in this condition are hamartomatous and rarely undergo malignant transition but a few such cases have been reported in literature [9].The patients are prone to many extra intestinal tumors like Testicular Sertoli tumors, Ovarian tumors like sex cord tumors with annular tubules, Granulose Theca cell tumors and cystadenomas, Breast tumors like breast carcinoma, papilloma with squamous metaplasia, Cholangioma, pancreatic adenocarcinoma, adenoma malignum, bronchial carcinoids, papillomas in bladder and pelvis. This patient is on regular follow up has no extraintestinal tumors till date.

Timely polypectomy, clinicohistological correlation and follow-up of gastrointestinal polyps is necessary along with lifelong screening of malignancies on a regular basis. It is also necessary to investigate all first-degree relatives of the patient. Although the incidence of PJS is low, it is important for clinicians to recognize these disorders and to perform presymptomatic testing in patients at risk to prevent morbidity and mortality. [10]

ISSN: 0975-3583, 0976-2833 VOL14, ISSUE 09, 2023

Fig 1. Gross pathology of jejuno-jejunal intussusception

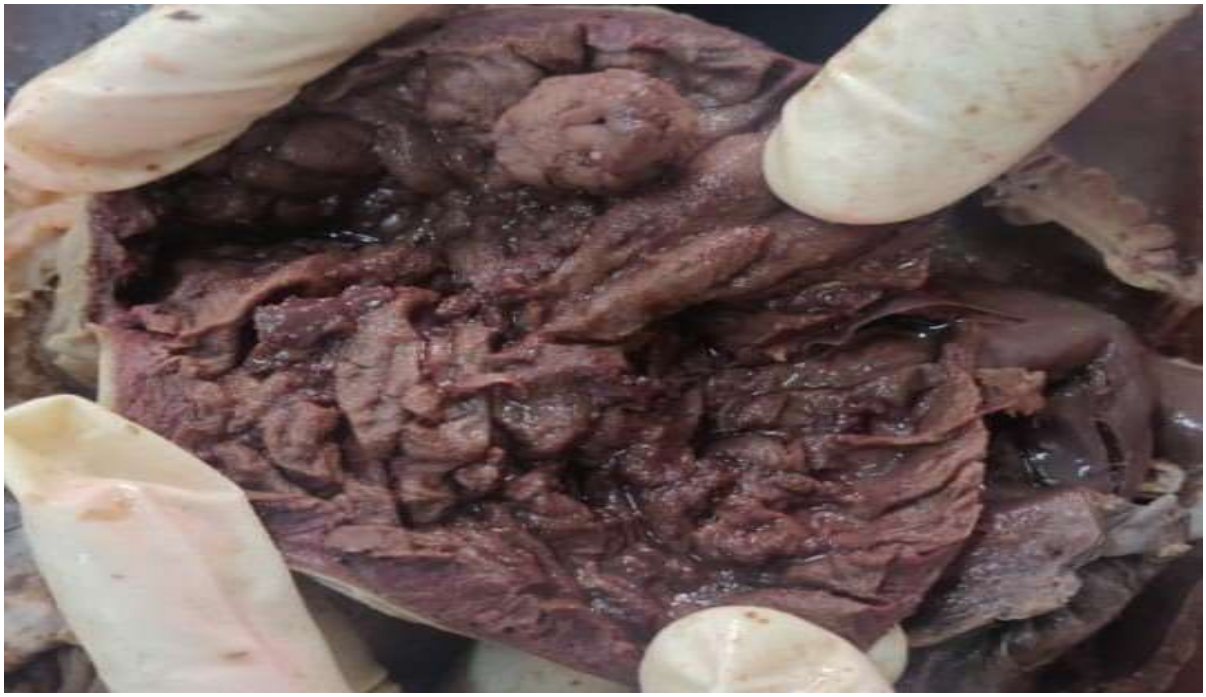

Fig 2. Pedunculated polyp inside the jejunum

ISSN: 0975-3583, 0976-2833 VOL14, ISSUE 09, 2023

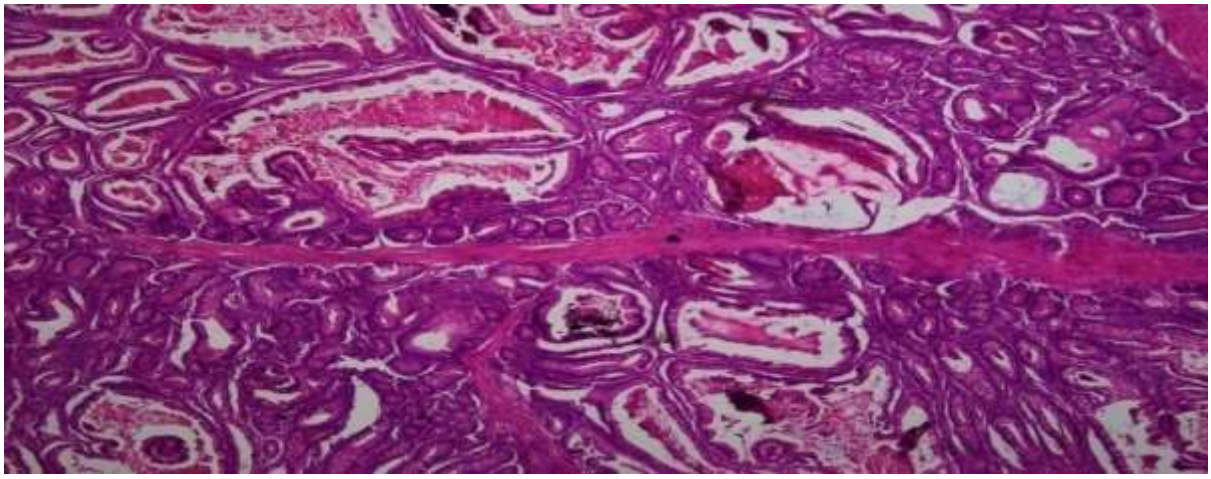

Fig 3. Polyp showing characteristic arborising network of connective tissue, bands of smooth muscle intermixed with lamina propria (H&E, x400)

Fig 4. Mucocutaneous pigmentation

CONCLUSION

Peutz- Jeghers syndrome is a rare autosomal dominant disorder which can often lead to intestinal intussusception due to gastrointestinal polyps. The prognosis of the patient can be improved by timely clinical diagnosis with histological correlation. Due to its high gastrointestinal and non gastrointestinal malignant potential, it is recommended to keep the patient under lifelong monitoring and essential screening of first degree relatives.

BIBLIOGRAPHY

1.Beggs AD, Latchford AR, Vasen HFA, Moslein G, Alonso A, Aretz S, et al. Peutz-Jeghers syndrome: a systematic review and recommendations for management. Gut 59 (2010): 975-86.

2. Santosh T, Patro MK, Nayak J and Behera B. A classical case of Peutz–Jeghers syndrome with brief review of literature. Human Pathology: Case Reports 4 (2016): 9-12.

ISSN: 0975-3583, 0976-2833 VOL14, ISSUE 09, 2023

3. Algarni M, Raml E, Trabulsi N and Nassif M. Small bowel intussusception and concurrent sigmoid polyp with malignant transformation in Peutz–Jeghers syndrome, J Surg Case Rep. 2019Jan; 2019(1).

4. 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

5.Tse J, Wu S, Shinagare S, Lauwers GY, Yilmaz O,Wu CL, et al. Peutz-Jeghers syndrome: a critical look at colonic Peutz-Jeghers polyps. Mod Pathol.2013 Sep;26(9):1235-40

6. Giardiello FM, Brensinger JD, Tersmette AC, Goodman SN, Petersen GM, Booker SV, et al. Very high risk of cancer in familial Peutz-Jeghers syndrome. J Gasteroenterol. 2000 Dec;119(6):1447-53.

7. Jenne DE, Reimann H, Nezu J, Friedel W, Loff S, Jeschke R, et al. Peutz-Jeghers syndrome is caused by mutations in a novel serine threonine. Nat Genet 1998;18(1):38-43.

8. Hemminki A, Markie D, Tomlinson I, Avizienyte E, Roth S, Loukola A, et al. A serine/threonine kinase gene defective in Peutz-Jeghers syndrome. Nature 1998;391(6663):184-7.

9. Mehta MV, Porecha MM, Mehta PJ: Small intestinal adenocarcinoma in Peutz-Jeghers syndrome. Indian J Gastroenterol 2006,25:38-39.

10. Kopacova M, Tacheci I, Rejchrt S, Bures J. Peutz-Jeghers syndrome: diagnostic and therapeutic approach. World journal of gastroenterology: WJG. 2009 Nov 21;15(43):5397.