Case Report

DOI: NJLM/2013/6968:1984

Pathology Section

Solitary Peutz Jeghers Polyp Causing Jejunal-Jejunal Intussusception in 6-Year-Old Female Child

KHUSHBOO BIRLA, SUNITA SANJAY DANTKALE, HEMLATA KAMRA, PANKAJ W. SAKINLAWAR, SWAPNIL D. CHANDEKAR

ABSTRACT

Peutz-Jeghers Syndrome (PJS) is a rare autosomal dominant syndrome characterised by the familial occurrence of gastrointestinal hamartomatous polyps in association with mucocutaneous hyperpigmentation. We here report a case of 6 yr old female child presented with colicky abdominal pain, nausea and vomiting. The patient underwent laparotomy due to small bowel intussusception secondary to a jejunal polyp. Histological examination showed the characteristic features of Peutz-Jeghers Polyp (PJP), but no other features of (PJS) as laid down by WHO were seen. Solitary PJP is an extremely rare condition, with an estimated incidence of 1:1,20,000 and all previously reported patients were older than this patient.

Key Words: Peutz-Jeghers Syndrome, Solitary Peutz-Jeghers Polyp, Hamartomatous, Gene, STK-11, Intussusception

INTRODUCTION

Peutz-Jeghers Syndrome (PJS) was first described by Peutz in 1921 and subsequently elaborated upon by Jeghers in 1949 [1]. Other designations used synonymously, includes periorificial lentiginosis/polyps and spots syndrome. It is seen in both male and female patients with no racial predominance [2]. WHO has laid down the following criteria for diagnosis of Peutz-Jeghers Syndrome: i) Three or more histologically confirmed Peutz-Jeghers polyps or ii) Any number of Peutz-Jeghers Polyp with a family history of Peutz-Jeghers Syndrome or iii) Characteristic, prominent, mucocutaneous pigmentation with a family history of Peutz-Jeghers Syndrome or iv) Any number of Peutz-Jeghers Polyp and characteristic, prominent mucocutaneous pigmentation [3].

Solitary Peutz-Jeghers Polyp (PJPs) occurs rarely, [4] though cases have been reported in the literature [5]. It is still unclear whether solitary PJP represents an incomplete form of PJS or a different entity [6].

We here report a case of solitary PJP in a 6 yr old female child, in whom the other features of PJS were lacking.

CASE PRESENTATION

A 6 yr old female child presented to the emergency department with colicky and intermittent abdominal pain since 1 day. Patient also had complains of nausea and several episodes of non-bilious, non-bloody vomiting for last 3 days and on & off constipation since 1 year. Abdominal examination revealed tenderness and guarding over left lumbar and periumblical region along with palpable mass of 15x10 cm in the same region. Blood counts were within normal limits. X-ray abdomen revealed target sign and on abdominal ultrasonography pseudokidney sign was noted, both consistent with intussusception. On emergency exploratory laparotomy, a jejunojejunal intussusception was seen located approximately 60 cm distal to the duodenojejunal flexure [Table/Fig-1]. Following careful manual reduction, a single, soft, sessile polypoidal mass 3 cm in diameter was noted forming leading edge of intussusceptions [Table/Fig-2 and 3]. The lead-point polyp was resected along with a short segment of adjacent jejunum on either side, followed by jejunojejunostomy. Resected specimen was sent for histopathological examination. Intra-operatively whole gastrointestinal tract was screened for evidence of polyp at any other place.

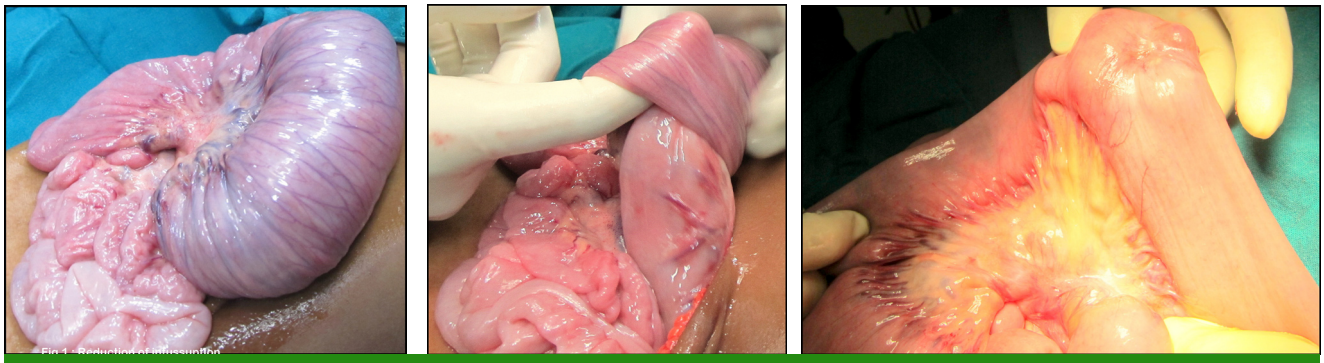
Gross specimen comprised of already cut open portion of jejunum measuring 11 cm in length that revealed a 4x3.5x2 cm sessile polypoidal growth [Table/Fig-4]. Externally there were pin point haemorrhages and cut surface was gray white solid homogenous [Table/Fig-5].

Histopathology showed a polyp composed of glandular epithelium resembling normal jejunal mucosa resting on central core of arborizing smooth muscle [Table/Fig-6-8]. Ischemic necrosis with haemorrhage was prominent over surface of the polyp, consistent with the leading edge of intussusception. Stroma showed inflammatory cell infiltrate.

Immunohistochemical stain of smooth muscle actin in the mucosal layer revealed a proliferation of smooth muscle bundles [Table/Fig-9 and 10]. No signs of dysplasia were seen and the surrounding tissue had a normal histological appearance. A diagnosis of Peutz Jeghers polyp was made. Patient was re-evaluated for other features suggesting PJS, but no other symptoms or signs of PJS were found like family history and pigmentation in oral cavity, around lips or orbital cavity.

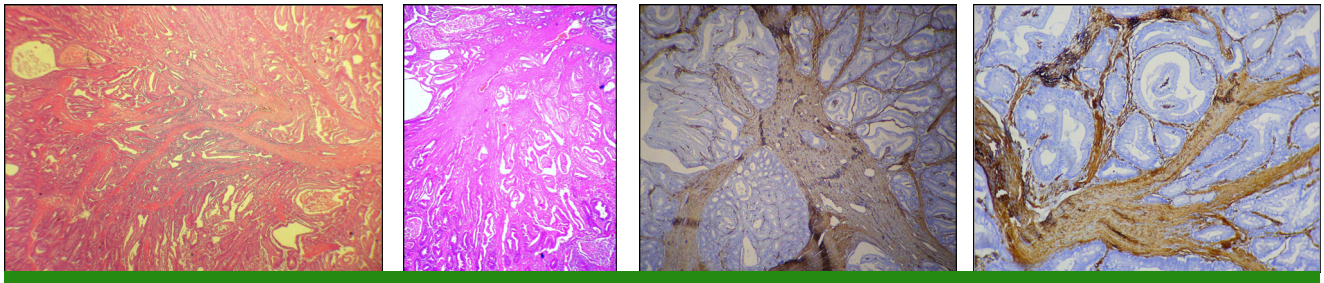
DISCUSSION

Hamartomatous polyps in the GI tract are a frequent cause of intussusception and are found in nearly all patients with Peutz-Jeghers Syndrome. [7] A hamartomatous polyp is diagnosed as a solitary Peutz-Jeghers polyp (PJP), when it doesn't suffice WHO criteria of PJS. Some studies document PJS being more common in 2nd or 3rd decade, and solitary PJP in 4th decade [8]. The youngest patient reported is a 13-year-old girl [9]. In 50–94% of patients with PJS, a mutation of



[Table/Fig-1]: Distended loops of jejunum due to intestinal obstruction. (1874x1630 pixels)
[Table/Fig-2]: Intraoperative manual reduction of intussusception. (1896x1850 pixels)
[Table/Fig-3]: Lead point polyp along with hemorrhagic bowel loops.(2896x1950pixels)

[Table/Fig-4]: Gross picture - Cut segment of jejunum with sessile polypoidal growth. (3130x3240 pixels)
[Table/Fig-5]: Gross picture - Cut surface of polyp: solid homogenous and tree like branching pattern. (3560x2680 pixels)
[Table/Fig-6]: M/E - Intestinal glands resting on central core of smooth muscle. (2650x2480 pixels) (H&E stain)



[Table/Fig-7, 8]: M/E - Ramifying bundles of smooth muscles in between glands. (2440x3080 pixels) (H&E stain)
[Table/Fig-9, 10]: M/E - Immunohistochemistry positive for smooth muscle actin. (4100x3460 pixels) (SMA)

the LKB1/ STK11 gene is found but some reports document absence of such mutation in solitary PJP [3].

Polyps of PJS are commonly found in the small intestine, but they can occur anywhere from stomach to rectum and other extra-intestinal sites like kidney, ureter, gallbladder, bronchial tree, and nasal passages [10]. The presenting gastrointestinal symptom in a series of patients with PJS was bleeding in 14%, intestinal obstruction in 43%, pain in 23% and extrusion of a rectal polyp in 7% of patients [11].

Typical imaging features of Peutz-Jeghers syndrome consist of multiple polypoid lesions variable in appearance—small or large, sessile or pedunculated. Apart from PJP multiple gastrointestinal polyps can be seen with other polyposis syndromes, such as familial adenomatous polyposis, juvenile polyposis, Cowden disease, and Cronkhite-Canada syndrome. Clinical features in conjunction with radiologic findings and histologic specimens help in correct diagnosis. Another important imaging finding in PJS is intussusception which is demonstrated as target and meniscus signs on abdominal X-ray. Ultrasonography and CT scan reveals “doughnut” or “pseudokidney” appearance for the same. On histology the most characteristic feature of a Peutz-Jeghers polyp is a central core of smooth muscle that extends into the polyp in an arborizing fashion and that is covered by either normal or hyperplastic mucosa native to the involved site [5].

The mucocutaneous lesions often seen on lips, buccal mucosa, perioral, and periorbital region are considered to be hamartomatous in origin and without potential of becoming malignant. Hamartomatous PJP are generally considered to have very low malignant potential. However, areas of neoplastic change have also been described in 3%-6% of the polyps in PJS [12]. Ichiyoshi et al., and Suzuki S et al., in two separate studies found evidence of focus of neoplasia in solitary PJP [13].

Patients with Peutz-Jeghers syndrome are at increased risk of developing gastrointestinal adenocarcinomas, an estimated frequency is approximately 2%–3% with the mean age at diagnosis being about 40 years of age. The majority of cancers are found in the stomach, duodenum, and colon. Small bowel is the least common site of adenocarcinoma [5].

Patients with Peutz-Jeghers syndrome are also at increased risk for developing extraintestinal malignancies, most common of which are pancreatic, breast, and reproductive organ carcinomas such as ovarian and testicular cancer. Other rare disorders, such as Sertoli cell tumour of the testis and sex cord tumours with annular tubules occur relatively quite frequent in patients with PJS, however none of these have been reported in patients with solitary PJP’s [3]. An upper gastrointestinal endoscopy and colonoscopy is necessary to rule out presence of hamartomatous polyps at other sites, a feature favouring PJS [9].

Intussusception is a common cause of abdominal emergency and intestinal obstruction in childhood. Approximately 90% of all paediatric intussusceptions are ileocolic, ileocecal. [14] Small bowel intussusceptions are much less common representing 1%-10% of all intussusceptions in younger children. Intussusception in an adult is often associated with a malignant lead point, in contrast to childhood intussusceptions which is often traced to a benign cause such as enlarged lymphoid tissue due to viral infection [15]. Specific lead points identified include Meckel’s diverticulum (75%) carcinoid tumours and leiomyoma, Peutz-Jeghers syndrome (16%), cystic fibrosis, ascaris infection, and other malignancies. After age of 14, only 10% of cases are considered idiopathic.

CONCLUSION

To conclude with, we want to emphasise that Intussusception is the second most common abdominal emergency after acute appendicitis in infants and children, and though it usually occur in absence of predisposing cause but a rare benign etiology of solitary PJP should also be kept in mind. Secondly, we here described a patient with a large, solitary PJP in the jejunum at an age typical of patients with PJS, but considerably younger than previously reported patients with solitary PJP’s.

The gene in Peutz-Jeghers syndrome appears to control growth and differentiation in the gastrointestinal tract, as shown by the development of hamartomas. In addition, there is clearly some evidence for a hamartoma/carcinoma sequence, and this has already been suggested for patients with juvenile polyposis. The gene is also expressed in other tissues, as shown by the abnormal freckling. The generally increased risk of cancer at a number of other sites suggests that the gene locus involved may be of importance to cancer development in general.

ACKNOWLEDGEMENT

1. I wish to express my profound gratitude and thanks to my teachers Dr. Sunita S. Dantkale and Dr. H.T. Kamra for the patient and dedicated supervision throughout the study.
2. A depth of gratitude also goes to my colleagues Dr Swapnil D. Chandekar and Dr Pankaj W. Sakinlawar for the comments and assessment, playing a positive and critical role in this project.

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AUTHOR(S):
1. Dr. Khushboo Birla
2. Dr. Sunita Sanjay Dantkale
3. Dr. Hemlata Kamra
4. Dr. Pankaj W. Sakinlawar
5. Dr. Swapnil D. Chandekar

PARTICULARS OF CONTRIBUTORS:
1. Post Graduate Student, Govt. Medical College and Hospital, Latur, Maharashtra, India.
2. Associate Professor, Govt. Medical College and Hospital, Latur, Maharashtra, India.
3. Associate Professor, Govt. Medical College and Hospital, Latur, Maharashtra, India.
4. Post Graduate Student, Govt. Medical College and Hospital, Latur, Maharashtra, India.
5. Post Graduate Student, Govt. Medical College and Hospital, Latur, Maharashtra, India.

NAME, ADDRESS, E-MAIL ID OF THE CORRESPONDING AUTHOR:
Dr. Khushboo Birla,
C/o Kishan Birla, Plot No. 88, 2nd A Road,
Sardarpura, Jodhpur-342001 Rajasthan, India.
Email: khushboobirla@gmail.com
Ph: 7303069592

FINANCIAL OR OTHER COMPETING INTERESTS: None.

Date of Publishing: **Aug 30, 2013**