Peutz Jegher's Syndrome - Double Intussusception in a Teenage Patient in a Single Setting - A Rare Case Report

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Published on 15th April 2024

ABSTRACT

Peutz jegher's syndrome is a rare autosomal dominant disorder with hamartomatous polyps and mucocutaneous melanin pigmentation. This case report shows a case of a teenage Indian boy presented with intestinal obstruction due to double intussusception in a single setting due to the PJS polyps. This is the first case of double intussusception in a teenage patient with PJS in a single setting reported in India and fifth case in the world. A 14-year-old teenage boy presented to a surgical emergency with complaints of abdominal pain, bloody stools, and vomiting with characteristic ink black macules over buccal mucosa. On abdominal examination, sausage-shaped mass was palpable and he was provisionally diagnosed with Peutz-Jeghers Syndrome. Ultrasound abdomen findings favoured the diagnosis of intussusception at two sites. He underwent Emergency Laparotomy with resection & anastomosis of involved segments of the ileum and jejunum. Intraoperative findings showed Jejunojejunal intussusception 50 cm distal to DJ junction with 25 cm of gangrenous jejunum and ileoileal intussusception 30 cm proximal to ileocaecal junction which was histopathologically found to be hamartomatous polyps. The postoperative period was uneventful. He is under surveillance. Periodic surveillance is helpful in patients with PJS.

Keywords: Peutz Jegher's Syndrome, Intussusception

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INTRODUCTION

Intussusception is the telescoping of the proximal segment of the intestine (intussusceptum) into a distal segment. Peutz-Jeghers syndrome is an autosomal dominant disorder with STK11 gene mutation in chromosome 19.[1] It leads to hamartomatous polyps as well as they possess an increased risk of multiple cancers. A common presentation of PJS is abdominal pain due to intussusception. Other patients usually present with one of the complications such as features of gastrointestinal bleeding (anemia, hematochezia, or hematemesis), intestinal obstruction, biliary obstruction, and gastric outlet obstruction.[2] Regular surveillance plays a pivotal role in preventing complications following intestinal obstruction due to intussusception in these PJS and early diagnosis of malignancy. It is diagnosed with histopathological determination of hamartomatous polyps and at least two clinical features including family history, two or more polyps, and mucocutaneous pigmentation. Here is a rare case report of a teenage boy presented to the emergency surgery casualty with double intussusception

CASE REPORT

A 14-year-old boy from Trivandrum presented to surgical casualty with complaints of acute abdominal pain distension and non-bilious vomiting for 1 day. It was associated with blood in stools on 22/10/22. On physical examination, he had tachycardia, pallor and multiple ink black macules over the buccal mucosa **(Figure 1).** On examination of the abdomen, he had a sausage-shaped mass in the right iliac fossa with guarding, rigidity as well as decreased bowel sounds. Per rectal examination showed ballooning and blood staining with no stools.

He was investigated with USG abdomen which showed bowel within bowel appearance in subhepatic region suggestive of intussusception with dilated small bowel loops in epigastric and left hypochondrium suggesting

Cite this article as: Devipriya NV, Sreekumar RC. Peutz Jegher's Syndrome - Double Intussusception in a Teenage Patient in a Single Setting - A Rare Case Report. Kerala Medical Journal. 2024 Apr 15;17(1):41–4. | DOI: https://doi.org/10.52314/kmj.2024.v17i1.630

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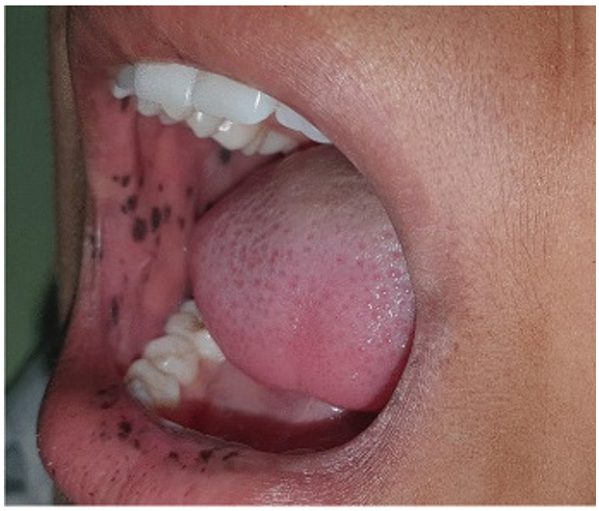

Figure 1. **Image showing ink black macules over buccal mucosa**

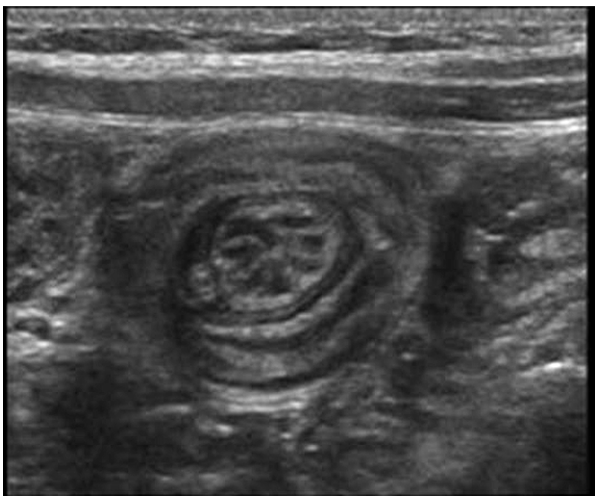

Figure 2. **Ultrasound image showing target sign**

small bowel obstruction **(Figure 2).**

He was provisionally diagnosed with Peutz-Jeghers syndrome based on clinical and radiological findings.

He underwent an emergency laparotomy on 22/10/22 under General anaesthesia. Intraoperatively we found double intussusception in a single setting firstly Jejuno-jejunal intussusception 50 cm distal to DJ junction with 25 cm of gangrenous jejunum, resection and end-end anastomosis of jejunum done. Secondly, ileoileal intussusception 30 cm proximal to ileocaecal junction, 10 cm of gangrenous ileum resected, then end-end anastomosis done. Besides, there was a single large 7 cm polyp in ileum. Jejunum had multiple small polyps with an enlarged mesenteric lymph node of size 1 x 1 cm size **(Figure 3, Figure 4, Figure 5).**

Postoperatively patient was started on orals, tolerated, and he was discharged after 1 week. He was followed up in our OPD and the histopathology report was consistent with hamartomatous polyps of Peutz Jeghers syndrome (containing arborising smooth muscles). He is under regular surveillance. His family members were also advised on periodic surveillance with imaging, endoscopy, and colonoscopy.

DISCUSSION

Peutz Jeghers syndrome belongs to a group of familial polyposis syndromes characterised by development of multiple hamartomatous polyps in gastrointestinal tract, mucocutaneous pigmentation and elevated risk of developing cancers caused by mutation of STK11 gene in chromosome 19p13. It has a prevalence of 1:100000 births with equal male and female preponderance. It was first reported in identical twins in 1895 by Dr.Connor, a British physician and one of the twins died of intussusception at the age of 20 and the other due to carcinoma breast at 59 years of age.[3] It was first described by Peutz in 1921 in a Dutch family with gastrointestinal polyps and mucocutaneous lesions. It was known as PJS since 1954.[4] Later, in 1998, it was discovered that the cause

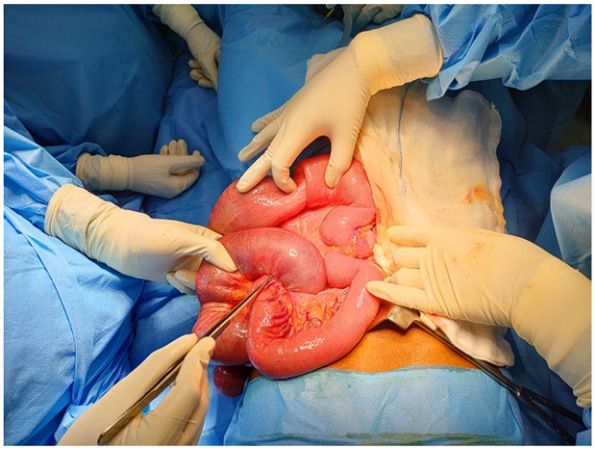

Figure 3. **Intraoperative image showing jejunojejunal and ileoileal intussusception**

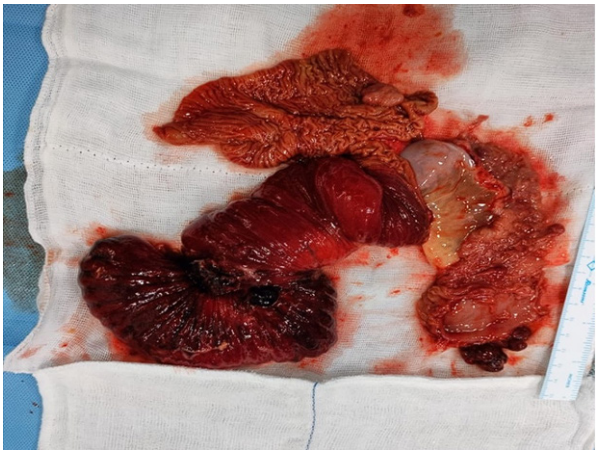

Figure 4. **Intraoperative image showing multiple polyps with gangrenous resected out jejunum**

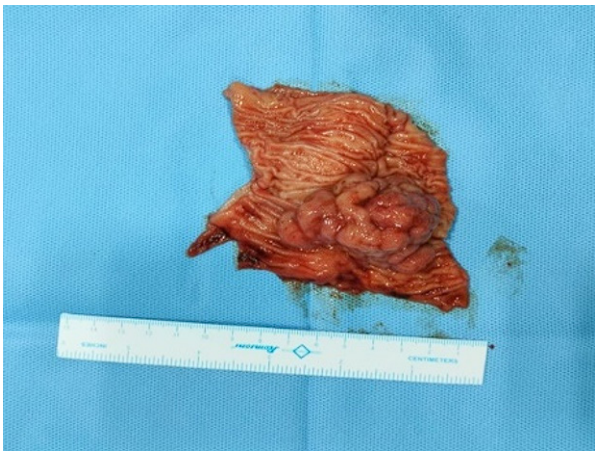

Figure 5. **Resected out specimen of ileum showing lead point polyp causing intussusception**

of PJS was a mutation in the STK11 gene, also known as the LKB1 gene.[5] STK11/LKB1 genetic mutation is found in around 30% to 70% of PJS patients and in around 70% of patients with a positive family history.[6] Beggs et al defined the diagnostic criteria of PJS as following when any one of the following is present:

- Two or more histologically confirmed PJ polyps.
- Any number of PJ polyps detected in one individual with a family history of PJS in a close relative
- Characteristic mucocutaneous pigmentation in an individual who has a family history of PJS in close relative.
- Any number of PJ polyps in an individual who also has characteristic mucocutaneous pigmentation.[7]

The classical triad for intussusception of abdominal pain, mass, and bloody stool was present in our patient. Patients with PJS generally have hundreds of polyposis and are mainly distributed in the small intestine, followed by the large intestine and stomach. Usually, the intestinal polyps are pedunculated, while the polyps in the stomach are broad-based and hamartomatous histologically.[8]

Baeza-Herrera et al.[9] in 2005, reported that 1.4% of children with PJS also had intussusception. Nowadays, conventional barium studies have been replaced with more advanced cross-sectional techniques such as ultrasonography, CT, and magnetic resonance imaging (MRI). Ultrasonography will typically show a "doughnut" or a "target" sign picture; however, in this patient, ultrasound was not performed. An abdominal CT scan is the most useful imaging modality as it helps reveal the underlying lesion.[10] Hence, patients with PJS are strongly recommended to have regular follow-up examinations and periodic polypectomy of the entire small bowel to prevent complications.[11]

NEED FOR GENETIC COUNSELLING IN PJS PATIENT

Genetic counselling is important in a PJS patient because it aids in averting the complications (eg. short gut syndrome) caused by the recurrent need of resections and in creating awareness about screening for PJS-associated cancers & thereby increasing the rate of early detection plus reducing associated mortality & morbidity.

Those who met the above criteria can be assumed to be having PJS. They should undergo genetic testing that includes a blood test to look for a mutation in the STK11 gene. If it is detected other family members should also undergo genetic testing. Genetic counselling is an inevitable part of the multidisciplinary management of Peutz Jegher's Syndrome. Another appreciable part of genetic counselling is that it allows a PJS parent to decide whether to carry his altered genes to offspring through **Prenatal Diagnosis** (Chorionic villous sampling/amniocentesis) and **Preimplantation Genetic Diagnosis (PGD).** In cases of IVF, this aids in **Negative Eugenics.**

CONCLUSION

Intussusception is common cause of acute abdomen in patients with Peutz Jeghers syndrome. Wireless capsule endoscopy is an emerging and popular diagnostic method and has been found to have a higher diagnostic yield of polyps as compared to a CT or MRI study.[12] Periodic regular surveillance is essential in PJS patients to avoid recurrent laparotomies and bowel resection leading to short gut syndrome which can affect their daily life. Prophylactic endoscopic polypectomy can also aid in preventing intussusception.[11] Surgical corrections pay a pivotal role in the management of Peutz Jegher's intussusception.

END NOTE

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Acknowledgement: The authors would like to express the gratitude towards Dr. Abdul Latheef (Prior

Head of Department of General Surgery, Government Medical College Thiruvananthapuram), Dr. Nizarudeen A (Present Head of Department of General Surgery, Government medical college Thiruvananthapuram), Dr. Anil Sundaram (Assistant Professor, Department of General Surgery, Government Medical College Thiruvananthapuram), Dr. Unnikrishnan (Assistant Professor, Department of General Surgery, Government Medical College Thiruvananthapuram), Dr. Rajesh (Assistant Professor, Department of General Surgery, Government Medical College, Thiruvananthapuram), and all Junior Residents in the department of General Surgery.

Funding & Conflict of Interest: The authors have not received any funding or benefits from industry or elsewhere to conduct this study.

Ethical Approval: Not required. Consent was taken from the patient for publishing data.

Data Availability: Data regarding this study is available with the corresponding author. Data will be provided at request.

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