

Case Report

Peutz Jeghers syndrome with multiple intussusceptions

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ABSTRACT

Peutz Jegher's syndrome is a rare autosomal dominant disorder characterized by the development of hamartomatous polyps and mucocutaneous melanin pigmentation. This case report shows the case of an adult Indian female with intestinal obstruction due to multiple intussusceptions that were caused by hamartomatous polyp of Peutz Jegher's syndrome. A 32-year-old female presented to the surgical emergency with complaints of colicky abdominal pain, vomiting and melena and with a positive family history for Peutz Jegher's syndrome. On abdominal examination, a mass was palpable in her right lumbar area and per rectal examination showed ballooning and blood staining. She was provisionally diagnosed with Peutz Jegher's syndrome with intussusception and her ultrasound imaging of the abdomen also showed the features suggestive of intussusception. She underwent an emergency laparotomy with resection and anastomosis of the involved segment of the jejunum and limited right hemicolectomy. Intraoperative findings showed multiple intussusceptions in her both large and small bowels along with multiple polyps which was seen to be hamartomatous polyps in the histopathological studies. The postoperative period was uneventful and during the review she was healthy. Intussusception is the major complication of Peutz Jegher's syndrome and it can be prevented with regular surveillance in known cases. Intussusception should be kept in mind as the first differential diagnosis when these patients present with acute abdomen. The investigation of choice that's preferred is a cross sectional imaging of the gastrointestinal tract to confirm with the diagnosis. Periodic surveillance is essential in those patients with known history or those with the mutation of STK11 (LKB1) gene.

Keywords: Peutz Jeghers syndrome, Intussusception, Hamartomatous polyps, Case report

INTRODUCTION

Peutz Jeghers syndrome has an autosomal dominant inheritance pattern and is caused by the mutation of STK11 (LKB1) gene. There are many mutations in the STK11 gene that was reported to be the cause.¹ It's diagnosed with histopathological determination of hamartomatous polyps and at least two of the clinical features including family history, two or more polyps, mucocutaneous hyperpigmentation of mouth, lips, noses, eyes, genitalia, or fingers.²

Patients have an increased chance of complications like intussusception that can lead to intestinal obstruction.³ They also have a predisposition to both gastrointestinal

and non-gastrointestinal malignancies.⁴ So active surveillance is essential in all cases. In every case, it is essential that early identification of Peutz Jeghers syndrome is necessary and that can reduce its complications and also help to screen them for cancers.⁵ Here we shed light upon a case of an adult female who presented with multiple intussusceptions in the emergency.

CASE REPORT

A 32-year-old female presented to the surgical emergency with complaints of recurrent episodes of colicky abdominal pain, vomiting of 3 months duration and aggravated for the last 3 days, and also melena of 4 months

duration. She did not have a history of any co-morbid illness in the past. Surgical history includes two lower segment caesarean sections in the past. Past medical history is significant for her father being diagnosed with Peutz Jeghers syndrome at the age of 51 years and underwent surgical management for the same from a tertiary care hospital. Neither he nor his family followed up with any surveillance for the disease.

On physical examination, she had pallor and mucocutaneous hyperpigmentation in the oral cavity (Figures 1 and 2). The abdominal examination revealed a firm non-tender mass of size 5×5 cm in the right lumbar area that appears and disappears simultaneously with pain. The digital per rectal examination showed ballooning and blood-stained stools.



Figure 1: Mucocutaneous hyperpigmentation in the buccal mucosa.



Figure 2: Mucocutaneous hyperpigmentation over the tongue.

Her blood investigations pointed to anaemia (Hb: 6 g/dl). The rest of the laboratory parameters were within normal limits. Imaging was carried out in the emergency setting with an ultrasound of the abdomen that depicted ileo-ileal intussusception. She was provisionally diagnosed with Peutz Jeghers syndrome with intussusception in view of the clinical and radiological findings and the positive family history. She was posted for emergency exploratory laparotomy.

She underwent an emergency midline laparotomy with resection and end-to-end anastomosis of the involved segment of the jejunum and limited right hemicolectomy. Intraoperative findings were significant for multiple intussusceptions (Figure 3). There were two jejuno-jejunal intussusceptions (Figure 3), they were located 15 cm and 35 cm respectively from the duodeno-jejunal flexure and the appendix was seen to be intussuscepted into the caecum (Figure 4) and all the three intussusceptions had a polyp as the leading point. There was also another ileo-cecal intussusception with a segment of ileum showing a polyp like growth located 40 cm from the ileo-cecal junction almost obstructing the lumen (Figures 5 and 6). The rest of the bowel had few small polyps in the lumen (Figure 7).



Figure 3: Intraoperative picture showing the multiple intussusceptions in the jejunum.

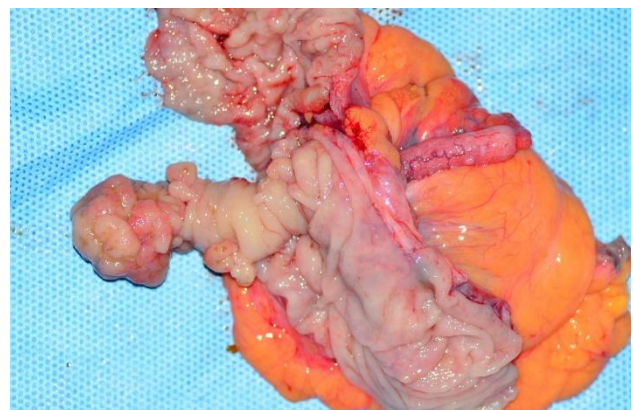


Figure 4: Specimen showing that the appendix was intussuscepted into the caecum with the polyp as the leading factor.

The postoperative period was uneventful. The histopathological examination of the specimen revealed hamartomatous polyps of the gastrointestinal tract. Hence, Peutz Jeghers syndrome was confirmed as the diagnosis. The patient was discharged home after one week and was asymptomatic at follow-up visit after one month. Aggressive screening was advised for the patient and the family including children and siblings.



Figure 5: Specimen showing the polyp at the ileum that caused the ileo-caecal intussusception.



Figure 6: The lumen obstructing large polyp.



Figure 7: Multiple small polyps in the lumen of ileum.

DISCUSSION

Peutz-Jeghers syndrome is an autosomal dominant condition characterized by the development of hamartomatous polyps in the gastrointestinal tract (stomach, small intestine, colon, rectum) and mucocutaneous pigmentation. It also has a propensity to cause solid organ malignancies of the pancreas, lung, breast, ovary, uterus and testes, apart from gastrointestinal neoplasms.

The prevalence of Peutz-Jeghers syndrome is estimated to be 1 in 100000 individuals.⁶ Peutz-Jeghers syndrome was first reported in identical twins in 1895 and one of the twins died of intussusception at age 20.⁷ It was first described by Peutz in 1921 in a Dutch family with gastrointestinal polyposis and pigmented lesions and known as PJS since 1954. Through this case report, we present a case of intussusception due to Peutz-Jeghers syndrome which is recognized as a common complication. But it is rare to see multiple intussusceptions in a single individual as in this case.

Clinically, Peutz-Jeghers syndrome patients present with hyperpigmented macules on lips, mouth, nostrils, buccal or intestinal mucosa which are present in 95% of patients, which are due to pigment-laden macrophages in the dermis.⁶ Patients may also have intermittent abdominal pain and the median time to onset for gastrointestinal symptoms or polypectomy is 13 years of age.^{8,9} The polyps are most frequent in the small bowel mainly the jejunum. Peutz-Jeghers syndrome is diagnosed with histopathological confirmation of hamartomatous polyp along with at least two of the following clinical criteria: positive family history, small intestinal polyps and mucosal hyperpigmentation. In histopathology, Peutz-Jeghers syndrome associated polyps demonstrate an arborizing pattern of smooth muscle proliferation.⁶

The complications of Peutz-Jeghers syndrome are intestinal obstruction due to intussusception, rectal bleeding, anemia and malignancy. In a study by Utsunomiya et al, intussusception was observed in 46.9% of patients.¹⁰

Genetic testing for STK11 gene on chromosome 19p13.3 can be done to identify Peutz-Jeghers syndrome.⁶ Diagnosis and treatment of this syndrome can be done by endoscopic removal of polyps larger than 5 mm using a polypectomy snare using intraoperative enteroscopy (IOE), conducted during laparotomy. Nowadays, it is replaced by double-balloon enteroscopy (DBE) in combination with capsule endoscopy. Complete polypectomy through IOE or DBE is conducive to longer symptom-free survival.¹¹ Perforation and bleeding are the anticipated complications of these endoscopic procedures.¹² Analogs of rapamycin which is a macrolide antibiotic are also used in clinical trials for the treatment of Peutz-Jeghers syndrome which could potentially lead to reduction in polyp burden and size.⁶ COX-2 inhibitors are also being evaluated for beneficial effect in reducing tumor burden and polyp size in the suffering patients.¹³

Periodic screening and timely polypectomy may obviate the need for surgical resection of the involved bowel.¹⁴ The risk of developing any malignancy in Peutz-Jeghers syndrome is 85% at age 70.¹⁵ The most common extraintestinal malignancy is breast cancer with a cumulative risk of 54%.¹⁶ For small bowel surveillance in Peutz-Jeghers syndrome, capsule endoscopy is used to detect small polyps while MRI enteroclysis is a safe

alternative for large-sized polyps.¹⁷ Screening for cancer of the colon, small intestine and oral cavity should begin at age 15 in Peutz-Jeghers syndrome patients. Screening for breast cancer is started at age 21 while malignancies of the thyroid, pancreas, ovary, testes, uterus and lung are screened from age 18.¹⁸

CONCLUSION

Intussusception is the major complication of Peutz-Jeghers syndrome and it can be prevented with regular surveillance in known cases. Intussusception should be kept in mind as the first differential diagnosis when these patients present with acute abdomen. The investigation of choice that's preferred is a cross-sectional imaging of the gastrointestinal tract to confirm with the diagnosis. Periodic surveillance is essential in those patients with known history or those with the mutation of STK11 (LKB1) gene.

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