

Peutz-Jehger's Syndrome presenting as adult intussusception

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Abstract: Peutz-Jehger's syndrome is an autosomal dominant disorder. It is the second most common hamartomatous syndrome with hamartomatous polyp of gastrointestinal tract and cutaneous melanin deposit. Majority of patients remain relatively asymptomatic. Some present with abdominal pain secondary to obstruction or impending obstruction owing to an intussuscepted polyp with GI bleeding. We report a case of 21 year male who presented with upper central abdominal pain, distension, vomiting and blood mixed stool. He also had black colored spots on perioral region and fingers. Ultrasound revealed mass lesion with echogenic centre suggestive of intussusception. Emergency laparotomy revealed jejunojejunal intussusception with gangrenous intussusceptum and two other large polyps. Resection anastomosis of jejunum and excision of polyps was done. Peutz-Jehger's syndrome is a rare but important cause of intussusception in adults presenting with cutaneous manifestations and features of intestinal obstruction.

Key words: Cutaneous melanin, Hamartomatous polyp, Intussusception, Peutz-Jegher's syndrome

Introduction

Peutz-Jeghers syndrome (PJS) is a rare autosomal dominant disease that is characterized by hamartomatous gastrointestinal polyps in association with pigmentation affecting skin and mucous membranes.¹ Anemia can result from chronic bleeding. Other mode of presentation is recurrent obstruction and intussusception.² Resection anastomosis of the involved segment and excision of palpable polyps larger than 1.5 cm is the treatment of choice.

Case Report

A 21 year male presented with central upper abdominal pain with distension. There was history of vomiting and blood mixed stool for 5 days. On examination, patient was pale dehydrated with perioral and submucous melanin deposits (Fig 1). Abdomen was slightly distended with periumbilical fullness and a vague mass which was "sausage" shaped and tender on touch. Per rectal examination revealed black coloured stool.



Fig. 1. Melanosis of digits and oral cavity

Peutz-Jehger's Syndrome

Ultrasound of abdomen showed mass lesion located in central abdomen. It had an echogenic centre and alternate hypoechoic and echogenic peripheral layers, giving appearance of a "target" in transverse scan and pseudokidney sign in longitudinal scan. The ultrasonographic impression was that of Intussusception (Ileo-ileal).

X-ray abdomen showed mild dilatation of jejunal loops with few air fluid levels.

Patient underwent emergency laparotomy and the findings were jejuno-jejunal intussusception approximately 65 cm distal to the duodenojejunal junction with small perforation in the intussusceptum (Fig. 2). There were two polyps of



Fig 2. Small bowel intussusception

4x4 cm, 20 cm and 45 cm distal to duodenojejunal junction. Resection of the part involved in intussusception and anastomosis of the adjacent healthy part was performed. Enterotomy and removal of the other two remaining polyps was also performed (Fig. 3).



Fig 3. Polyps seen after enterotomy

The postoperative recovery was uneventful. Histopathology of the resected part of small intestine and polyps revealed Gangrenous segment of small intestine with hyperplastic polyps (Fig 4). Lymph nodes showed non-caseating granulomatous and reactive lymphadenitis without any evidence of malignancy.

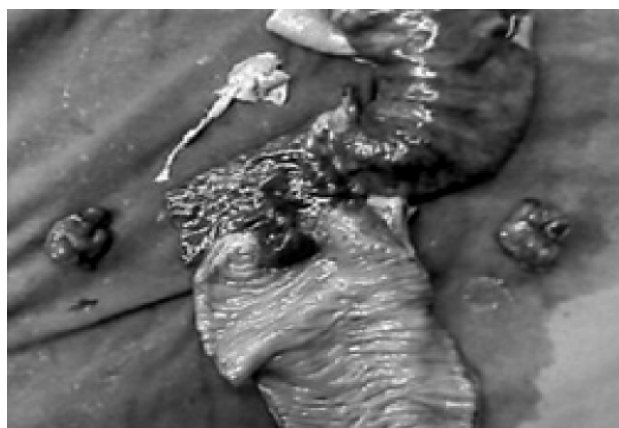


Fig 4. Resected specimen

Discussion

Peutz-Jeghers syndrome is due to germ line mutations in the Serine Threonine Kinase tumor-suppressor gene found on chromosome 19p13.3 (also called STK11/LKB1). The gene abnormality may be inherited or arise sporadically (35%). PJS has autosomal dominant inheritance. It affects males and females equally. Incidence is 1 in 30,000 to 120,000 live births ¹.

Clinical features of PJS can be divided into two types, cutaneous and gastrointestinal. The most noticeable cutaneous feature of PJS is the appearance of melanocytic macules (pigmented spots) in 95% of patients. Tan, dark brown, or bluish black flat patches 1 to 5 mm in size are seen around the mouth, lips, gums, inner lining of the mouth, eyes, hands and feet, fingers and toes, anus and genital areas. Pigmentation usually appears before 5 years of age and may fade after puberty. Gastrointestinal polyps occur later on in life and are rare in childhood. The polyps may cause bleeding and abdominal pain. ²

A polyp is a benign growth of the lining of the bowel. It can be anything from 2mm up to 5cm or more in diameter. Commonly, the abnormal cells form a small ball on the end of a stalk of normal cells. The type of cell that forms the polyp varies and is important in determining its potential for developing into a cancer.

Polyps usually cause no symptoms until they grow to 2cm or more in diameter. The most common symptom is rectal bleeding. Larger polyps can simulate faeces so the colon undergoes vigorous peristalsis in a futile attempt to expel the polyp which can lead to severe colicky pain.

Peutz-Jehgers polyps usually present in early adult life and carry a low but definite risk of malignancy, probably around five per cent per polyp, so they need excision. The number

of polyps per individual is variable and ranges, from as few as one - two to as many as 20 or more. Peutz-Jeghers-type hamartomatous polyps are most common in the small intestine (in order of prevalence: in the jejunum, ileum, and duodenum) but can also occur in the stomach and large bowel. They can result in chronic bleeding and anemia and cause recurrent obstruction and intussusceptions. Such polyps tend to present with symptoms of obstruction or abdominal pain. Diagnosis is usually made with barium X-rays and treatment generally is laparotomy with intussusceptions dealt in the standard manner.³

The age at onset for symptoms from polyps is variable, with some individuals developing symptoms within the first few years of life. In studies from MD Anderson Cancer Center, the median age at first GI symptoms was ten years, while the median age at first polypectomy was age 13 years.⁴ A report from Korea indicated a mean age of onset for GI symptoms of 12.5 years.⁵ In a review of 32 patients with PJS, laparotomy for bowel obstruction was performed in 30% of individuals by age ten years and in 68% by age 18 years.⁶

Routine endoscopic and intra operative enteroscopy with polypectomy decreases the frequency of emergency laparotomy and bowel loss resulting from intussusception.⁷ Laparotomy and intra operative endoscopies are appropriate for removal of polyps larger than 1.5 cm. Distal small-bowel polyps that are beyond the reach of conventional endoscopy have been difficult to manage. Until recently, barium contrast upper gastrointestinal series with a small-bowel follow-through has been recommended. However, Wireless capsule endoscopies⁸ and double-balloon, or "push and pull," enteroscopy⁹ can remove distal small-bowel polyps without laparotomy and allow better diagnosis and eradication of small-bowel polyps, often without laparotomy.

Peutz-Jeghers polyposis is associated with an increased risk of malignancy in the colon and small intestine as well as other extraintestinal malignancies like breast, cervical, ovarian, testicular, and pancreatic cancers¹. The risk of developing some form of internal cancer is 15 times greater for patients with PJS compared with the general population¹⁰. The John Hopkins University reported a 48% incidence of cancer, with 73% of tumors arising from gastrointestinal tract¹¹.

Conclusion

Peutz – Jegher's syndrome is a rare but important cause of adult intussusception. Though majority of the patients are relatively asymptomatic, abdominal pain and distension with cutaneous manifestations may guide towards the diagnosis.

References

1. Lindor MN, Greene MH. Peutz-Jeghers Syndrome (PJS). *Journal of the National Cancer Institute*. 1998; **90(14)**:1061-1062
2. Choi HS, Park YJ, Park JG. Peutz-Jehger's syndrome, a new understanding. *J Korean Med Sci* 1999; **14(1)**:2-7
3. McGarrity TJ, Kulin HE, Zaino RJ. Peutz-Jeghers syndrome. *Am J Gastroenterol* 2000; **95(3)**: 596-604.
4. Amos CI, Keitheri-Cheteri M, Sabripour M, McGarrity TJ, Seldin MF, Nations L et al: Genotype-phenotype correlation in Peutz-Jeghers syndrome *J Med Genet* . 2004; **41**:327-333
5. Choi HS, Park YJ, Youk EG, Yoon KA, Ku JL, Kim NK et al. Clinical characteristics of Peutz-Jeghers syndrome in Korean polyposis patients. *Int J Colorectal Dis*. 2000; **15**: 35–8
6. Hinds R, Philp C, Hyer W, Fell JM. Complications of childhood Peutz-Jeghers syndrome: implications for pediatric screening. *J Pediatr Gastroenterol Nutr*. 2004; **39**: 219–20.
7. Pennazio M, Rossini FP. Small bowel polyps in Peutz-Jeghers syndrome: management by combined push enteroscopy and intraoperative enteroscopy. *Gastrointest Endosc*. 2000; **51**: 304–8.
8. Parsi MA, Burke CA. Utility of capsule endoscopy in Peutz-Jeghers syndrome. *Gastrointest Endosc Clin N Am*. 2004; **14**: 159–67.
9. Ohmiya N, Taguchi A, Shirai K, Mabuchi N, Arakawa D, Kanazawa H et al. Endoscopic resection of Peutz-Jeghers polyps throughout the small intestine at double-balloon enteroscopy without laparotomy. *Gastrointest Endosc*. 2005; **61**: 140–7
10. Giardiello FM, Offerhaus JG. Phenotype and cancer risk of various polyposis syndromes. *Eur J Cancer* 1995; **31**: 1085-87.
11. Fenoglio-Preiser C. Polyposis syndromes. In Fenoglio-Preiser C, Lantz PE, Listrone MB, Davis M, Riike FO. *Gastrointestinal pathology. An atlas and text*. 2ed. Philadelphia: Lippincott-Raven publishers; 1999pp. 717-45