

Peutz-Jeghers syndrome presenting with small bowel obstruction as an emergency

A Kumar, P Shetty, B Anand Rao, Ramachandra

Citation

A Kumar, P Shetty, B Anand Rao, Ramachandra. *Peutz-Jeghers syndrome presenting with small bowel obstruction as an emergency*. The Internet Journal of Surgery. 2007 Volume 16 Number 2.

Abstract

Peutz-Jeghers syndrome (PJS) is an autosomal dominant hereditary disorder. Giardello et al. proposed diagnostic criteria for PJS: The definition 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.³ It is now acknowledged that patients with PJS are at higher risk of developing intestinal obstruction due to large polyps. Here we present a case of intestinal intussusception leading to obstruction in a patient with Peutz-Jeghers syndrome.

INTRODUCTION

Peutz-Jeghers syndrome (PJS) is an autosomal dominant hereditary disorder. It named after Dr. Johannes Peutz and Dr. Harold Jeghers who described a relation between mucocutaneous pigmentation and intestinal polyposis.^{1,2} The diagnostic criteria are histological confirmation of hamartomatous gastrointestinal polyps along with two of three of the below conditions: small bowel polyps, a family history of PJS and pigmented macules on the buccal mucosa, lips or digits.³ It is now acknowledged that patients with PJS are at higher risk of developing intestinal obstruction due to large polyps. Here we present a case of intestinal intussusception leading to obstruction in a patient with Peutz-Jeghers syndrome.

CASE REPORT

A 21-year-old female was referred to the department of surgery as an emergency with the complaints of abdominal pain and bilious vomiting for the past 14 days. On general examination, mucocutaneous melanotic macules were noted. On abdominal examination, there was a palpable mass, firm in consistency and tender, in the left hypochondrium. Rigidity was also noted. With these complaints the patient underwent emergency exploratory laparotomy without any radiological investigations being done. On table, intussusception of the bowel was seen. A firm mass was palpable in the lumen of the bowel. A segmental resection was done as the mass was completely obstructing the lumen and a polypoidal mass was found. It was sent for histopathological examination. A diagnosis of Peutz-Jeghers

syndrome was made clinically. Histopathological examination showed the hamartomatous nature of the polyps, strengthening our diagnosis. A history of similar mucocutaneous melanotic patches is noted in the family.

Figure 1

Figure 1: mucocutaneous melanotic lesions



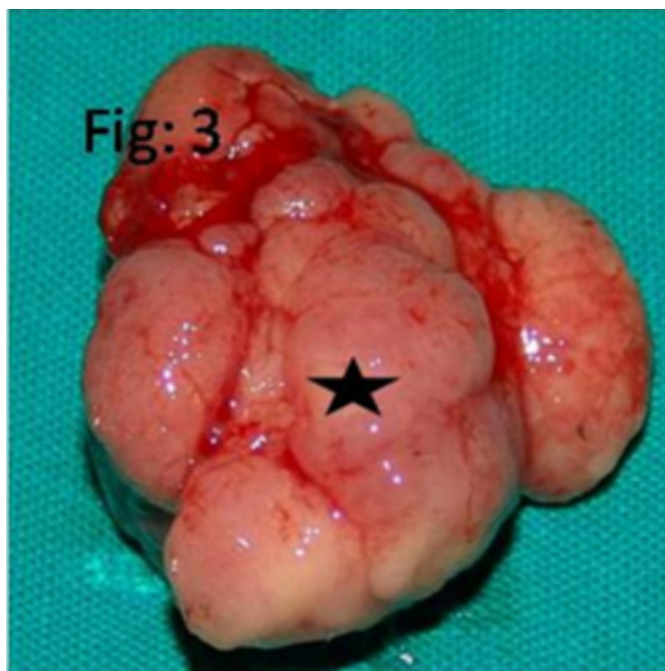
Figure 2

Figure 2: obstructed bowel due to intussusception



Figure 3

Figure 3: offending polyp that caused intussusception and obstruction



DISCUSSION

Peutz-Jeghers syndrome is an autosomal dominant hereditary disorder characterized by intestinal hamartomatous polyps and mucocutaneous melanocytic macules with a reported incidence of one in 83004 to one in 120 000⁵. Giardello et al. proposed diagnostic criteria for PJS³: The definition 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.^{3,6} Though mainly hamartomatous, few polyps may have

adenomatous components. Histologic examination of a polyp reveals a muscular core that extends in an arborial fashion into the superficial epithelial layer. Pigmented lesions are found in over 90 per cent of patients⁷, affecting the buccal mucosa, lips and digits, rectum, feet, vulva and conjunctiva. The causative locus for PJS is on chromosome 19p13.39. The gene is identified as LKB1 (also known as STK11), which encodes a serine-threonine protein kinase; a tumor suppressor gene.^{8,9} A possible second PJS locus at 19p13.4¹⁰ is suggested making Peutz-Jeghers syndrome genetically heterogenic. It is now universally accepted that patients with PJS are at higher risk of cancer than normal^{11,12}. The most common complications seem to be intestinal obstruction and anaemia. As the polyps can develop at any region in the entire GI tract, their recurrence is quite common, making it a challenging issue for the surgeon to make a decision regarding the amount of resections that can be done. Short-gut syndrome is a well known complication of repeated resections making the patient quite unstable due to malnourishment. In addition to that, a surgeon should also consider the potential for the development of intestinal adhesions with each surgery. Potential methods of investigating a patient with Peutz-Jeghers syndrome are stool examination for occult blood, gastrointestinal series, esophagogastroduodenoscopy, colonoscopy and CT scan.

Our patient, as she had already presented with intestinal obstruction and abdominal signs of peritonitis, underwent emergency laparotomy without any radiological investigation being undertaken. Intra-operatively, a huge polyp was identified as the culprit for intussusception leading to obstruction. This polyp was then removed by segmental resection of the bowel after reducing the intussusception. Post-operatively, the patient recovered well and she is on regular follow-up by GI endoscopy.

CORRESPONDENCE TO

Dr. Prashanth Shetty, M.B., B.S, M.S, Assistant Professor, Department of Surgery, Kasturba Medical College and Hospital, Manipal University, Manipal – 576104 Udupi District, Karnataka, India. Phone no.: +91 820 29 222 13 E-mail: drprashanthvshetty@yahoo.co.in

References

1. Peutz JL. Very remarkable case of familial polyposis of mucous membrane of intestinal tract and nasopharynx accompanied by peculiar pigmentation of skin and mucous membrane. *Nederl Maandisch Geneesk* 1921; 10: 134-146.
2. Jeghers H. Pigmentation of the skin. *N Engl JMed* 1944; 231: 88-100.
3. Giardello FM, Welsh SB, et al. Increased risk of cancer

in the Peutz-Jeghers syndrome. *N Engl J Med* 1987; 316: 1511-1514.

4. Mallery SB, Stough DB IV. Genodermatoses with malignant potential. *Dermatol Clin* 1987; 5: 221-230.

5. Hemminki A. The molecular basis and clinical aspects of Peutz-Jeghers syndrome. *Cell Mol Life Sci* 1999; 55: 735-750.

6. Boardman LA, Thibodeau SN, et al. Increased risk for cancer in patients with the Peutz-Jeghers syndrome. *Ann Intern Med* 1998; 128: 896-899.

7. Tomlinson IP, Houlston RS. Peutz-Jeghers syndrome. *J Med Genet* 1997; 34: 1007-1011.

8. Hemminki A, Markie D, et al. A serine/threonine kinase gene defective in Peutz-Jeghers syndrome. *Nature* 1998; 391: 184-187.

9. Jenne DE, Reimann H, et al. Peutz-Jeghers syndrome is caused by mutations in a novel serine/threonine kinase. *Nat Genet* 1998; 18: 38-43.

10. Mehenni H, Gehrig C, et al. Loss of LKB1 kinase activity in Peutz-Jeghers syndrome and evidence for allelic and locus heterogeneity. *Am J Hum Genet* 1998; 63: 1641-1650.

11. Spigelman AD, Murday V, Phillips RK. Cancer and the Peutz-Jeghers syndrome. *Gut* 1989; 30: 1588-1590.

12. Giardiello FM, Brensinger JD, et al. Very high risk of cancer in familial Peutz-Jeghers syndrome. *Gastroenterology* 2000; 119: 1447-1453.

13. Holt RW. Prevention of intussusception in Peutz-Jeghers syndrome. *Dis Colon Rectum* 1979; 22: 274-275.

Author Information

Addala Pavan Kumar

Post-Graduate Student, Department of General Surgery, Kasturba Medical College

Prashanth Shetty

Assistant Professor, Department of General Surgery, Kasturba Medical College

B.H. Anand Rao

Professor, Department of General Surgery, Kasturba Medical College

Ramachandra

Professor, Department of General Surgery, Kasturba Medical College