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

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Citation

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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 hamatomatous 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

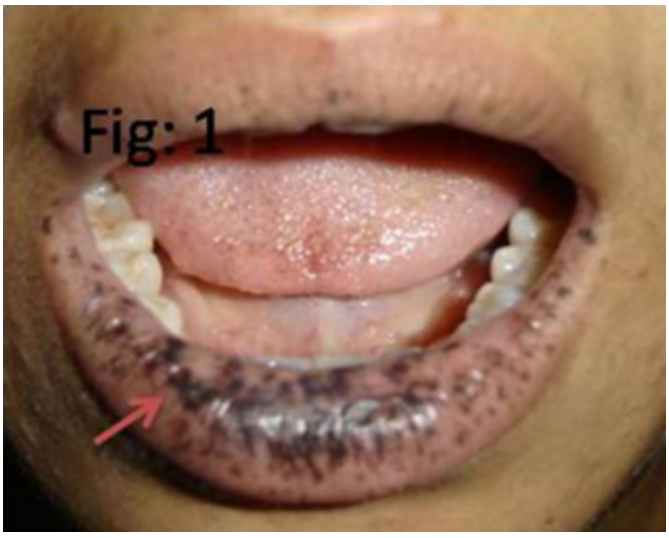


Figure 2

Figure 2: obstructed bowel due to intussusception

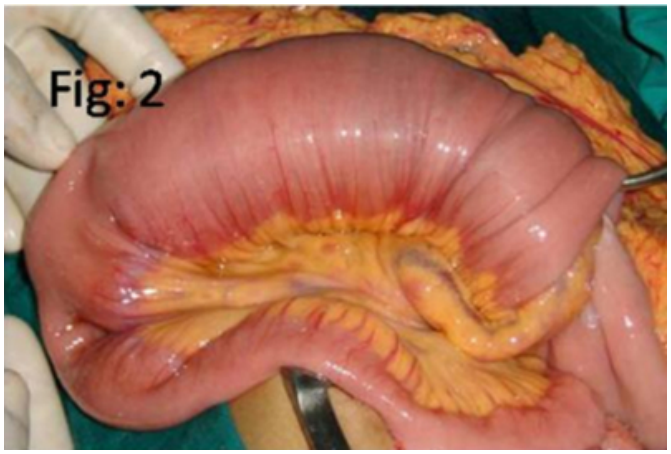
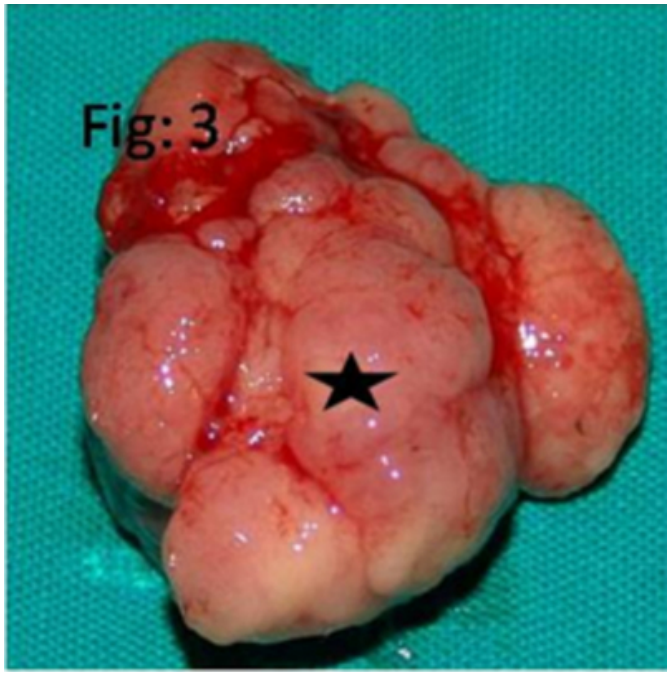


Figure 3

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 120 000[5]. 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 patients[7], 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 19p13.4[10] 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.

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