Case Report

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Surgery Section

Unusual Presentation of Intussusception of the Small Bowel with Peutz Jeghers Syndrome: Report of a Case

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ABSTRACT

The Peutz Jeghers syndrome (PJS) is an autosomal dominant disorder which is characterised by hamartomatous polyposes of the gastrointestinal tract, melanin pigmentation of the skin and mucous membranes, and an increased risk for cancer. We are reporting a case of a 15-year-old male with Peutz Jeghers syndrome, who presented to us with features of chronic intestinal obstruction and anaemia. Initially, patient was managed conservatively, but later on, an elective exploratory laparotomy was done for definitive management of intussusception. Laparotomy revealed a jejuno-jejunal intussusception with spontaneous recanalisation of gut which contained a long segment of gangrenous small bowel in the lumen. Resection and anastomosis of the jejunal segment was done. To the best of our knowledge, this might be the first case report on spontaneous recanalisation of small intestine.

Key Words: Peutz Jeghers syndrome, intussusception, spontaneous recanalisation, hamartomatous polyp, acute abdomen

INTRODUCTION

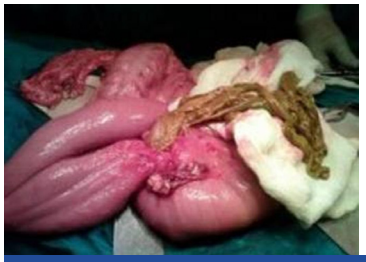
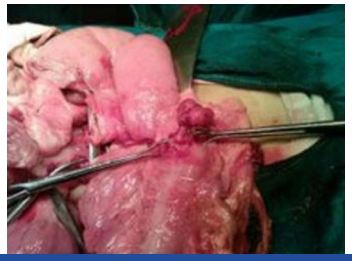
Peutz Jeghers Syndrome (PJS) is a complex hereditary polyposis condition. PJS is a rare disorder which is characterised by typical pigmented perioral macules, pigmented spots in the buccal mucosa which are present in 90% of patients, and the presence of multiple, although rarely more than twenty hamartomatous polyps, predominantly in the gastrointestinal tract. Polyps may occasionally be absent. Polyp sizes may vary from a few mm to 6 or 7 cm. Most of the patients have a characteristic clinical course of recurrent episodes of polyp induced bowel obstruction and bleeding. The disease affects males and females equally.

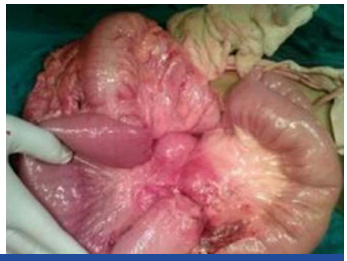
We report a case of 15-year-old male patient with PJS who presented to us with features of chronic intestinal obstruction and anemia. Patient was relieved of his symptoms related to intestinal obstruction by the conservative management but later on an elective exploratory laparotomy was done for persistent lower gastrointestinal bleed, on and off and anemia. Laparotomy revealed jejuno jejunal intussusception with spontaneous recanalisation of gut containing a long segment of gangrenous small bowel in the lumen [Table/Fig-1] and multiple polyps elsewhere in the gut [Table/Fig-2]. Since the mechanism of spontaneous recanalisation of the gut in this particular patient appeared to be unusual, we decided to report this with relevant review literature.

CASE REPORT

A 15-year-old male patient presented to us with complaints of abdominal pain and constipation which were there since one month. There was a past history of bleeding per rectum on and off since 3 years. There was a strong family history of PJS [Table/Fig-3]. Ten members of his family had PJS. His father, grandfather, two brothers, three cousin sisters and three uncles were found to be affected by the same disease. Three of them had died of the complications of the disease. Physical examination revealed pallor and melanin pigmentation over lips, buccal mucosa, hands and feet. The abdomen was soft, distended and tender, with no rigidity and no guarding. A movable firm lump which was of the size of a fist was felt in right hypochondrium. On per rectal examination, finger was stained with black coloured, soft faecal matter.

Patient's haemoglobin level was 5 g/dl and his renal and liver function tests were normal. Sigmoidoscopy revealed a single polyp which was about 22 cm from anal verge. Upper gastrointestinal endoscopy, ultrasonography of abdomen and barium enema studies showed no significant findings. CT scan and colonoscopy were not done because of unavailability of the same at our institute. Patient was kept on conservative management initially and a blood transfusion was done. An elective exploratory laparotomy was done 3 weeks later, for definitive management of intussusception and recurrent lower gastrointestinal bleeding. On exploration, a jejunojejunal intussusception was found [Table/Fig-4] and enterotomy revealed an approximately 30 cm long gangrenous small bowel segment which lay freely in the lumen and a large gangrenozed jejunal polyp (the lead point) at the apex of intussusception [Table/Fig-1]. Resection of the involved jejunal segment which was about 15 cm long was done



[Table/Fig-1]: Showing gangrenous intussusceptum coming out of enterotomy made in intussuscepted jejunal segment
[Table/Fig-2]: Showing colonic polyp visible through small colotomy incision
[Table/Fig-3]: Showing melanin pigmentations over lips of the father and the son with Peutz Jeghers syndrome
[Table/Fig-4]: Showing intussucepted segment of jejunu)

and the cut ends were anastomosed. One 5 cm size polyp of the transverse colon [Table/Fig-2] and another 3 cm size polyp of the sigmoid colon were removed through small colotomy incisions.

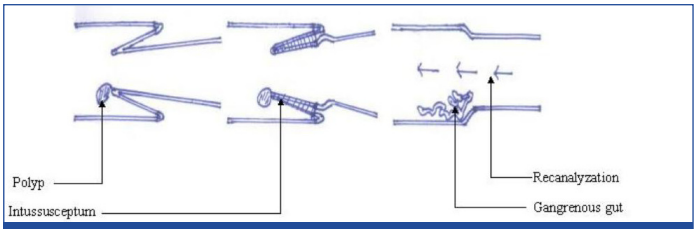
Histopathology examination (HPE) of the specimen showed necrotising inflammatory changes. HPE of the polyps was consistent with a diagnosis of hamartomatous polyps.

Postoperative recovery was uneventful and patient was found to be healthy at 2 months of follow up.

DISCUSSION

Peutz Jeghers syndrome is a complex hereditary polyposis condition which is characterised by autosomal dominant inheritance, hamartomatous polyps of the gastrointestinal tract, characteristic mucocutaneous pigmentation, and susceptibility to multiple cancers [1]. The cause of PJS appears to be a germline mutation of the STK11/LKB1 (serine/threonine kinase 11) tumour suppressor gene in most of the cases (70- 80%) [2, 3]. Its incidence has been estimated to be one in 120000 births [4]. The affected patients usually present with recurrent abdominal pain which is caused by intermittent intussusception or with manifestations of gastrointestinal bleeding. The sites which are most commonly affected by Peutz-Jeghers polyps in the gastrointestinal tract are the small bowel, colon, and stomach in decreasing frequency. The pedunculated nature of the polyps, combined with the large size to which the polyps can grow, can lead to recurrent intussusception in the small bowel [4]. There is an increased risk of gastrointestinal and non-gastrointestinal malignancies such as tumours of the breast, pancreas, lung, and reproductive tract in PJS patients. Although there is a genetic predisposition to cancers, a histological evidence of a hamartomatous-adenomatous-carcinomatous evolution and a direct hamartoma–carcinoma sequence in PJS patients have been documented in the literature. In addition, PJS patients can have both adenomatous and hamartomatous polyps separately, especially in the large intestine, and a malignant transformation of a small bowel hamartoma to a leiomyosarcoma has been reported [4]. The diagnostic criteria for PJS, which was proposed by Giardello et al., [5], includes histopathological confirmation of a hamartomatous gastrointestinal polyp and two of the following features: small bowel polyposis (at least two), a positive family history, and mucocutaneous hyperpigmentation.

As a non–invasive, inexpensive, and a readily available method, ultrasonography may demonstrate small bowel polyps in patients with Peutz Jeghers [6,7]. Ultrasonography may typically show a “doughnut” or a “target” sign [6] which is suggestive of an intussusception. A multicentric study showed that CT associated/not associated with barium enema may be the most accurate modality for diagnosis of adult intussusceptions [8]. A definitive surgical resection remains the recommended treatment in most of the cases of PJS which are associated with intussusceptions, because of the large proportions of structural causes and the relatively high incidence of malignancies [8]. In emergency surgical procedures, a combined endoscopic and surgical treatment is generally advocated, which includes IOE with endoscopic and/or surgical resection of polyps, with or without resection of short segments of intestine [9]. IOE is a combination of laparotomy with endoscopy,



[Table/Fig-5]: Showing schematic representation of probable process of gangrene of the intussusceptum with recanalisation of gut

which was accepted as the ultimate diagnostic and/or therapeutic procedure for complete investigation of the small intestine. Recent advances in genetic testing, magnetic resonance enterography, double-balloon endoscopy, IOE, and capsule endoscopy, all should result in an improved timely diagnosis and management of patients with PJS. Any polyp that is larger than 1.5 cm should be removed if possible, as it generally causes intussusceptions. It is reasonable to survey the colon endoscopically every 2 years and patients should be screened periodically for malignancies of GI tract [10].

In this case of Peutz Jeghers syndrome, recanalisation of the intussuscepted intestinal segment occurred spontaneously [Table/Fig-5] and the patient was able to tolerate food without much complaint.

To the best of our knowledge, this is the first report on spontaneous recanalisation of the small intestine in a case of chronic intussusceptions, where patient presented with chronic intestinal obstruction and anaemia. Probably this happened because of slow ischaemia which had allowed gangrene and subsequent detachment of intussusceptum and simultaneous spontaneous recanalisation of the gut [Table/Fig-5].

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