Intussusception of the small bowel due to Peutz-Jeghers syndrome: a case report

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Riassunto

La sindrome di Peutz-Jeghers è una malattia genetica autosomica dominante caratterizzata da una poliposi disseminata amartomatosa del tratto gastrointestinale, accompagnata da pigmentazione malanica della cute periorale e della bocca con un aumentato rischio di cancro. L'incidenza di complicanze chirurgiche in questi pazienti è abbastanza rara, e si correla alle dimensioni ed alla sede dei polipi. In questo lavoro, noi riportiamo un caso da noi osservato di una donna di 27 anni che si è presentata con dolori addominali, distensione addominale e vomito. Questa paziente presentava una caratteristica pigmentazione periorale. La diagnosi di occlusione intestinale da un polipo digiunale di grosse dimensioni e duplice intussuscezione è stata basata sui reperti del clisma del tenue e della TC e confermata alla laparotomia. Il trattamento eseguito (resezione limitata del tenue, riduzione dell'intussuscezione, enterotomie multiple con polipectomia), è risultato efficace nel controllo della complicanza, senza morbilita' associata.
Parole chiave: Sindrome di Peutz-Jeghers, intussuscezione digiunale.

Abstract

The Peutz-Jeghers syndrome is an autosomal dominant disorder characterized by hamartomatous polyposis of the gastrointestinal tract, melanin pigmentation of the skin and mucous membranes, and an increased risk for cancer. The incidence of surgical complications in these patients is relatively rare, and correlates with the size and location of the polyps. Herein we report the case of a 27-year-old woman presented with episodes of abdominal pain, abdominal distention and intermittent vomiting. Moreover, multiple pigmentation of the mouth was also noted. A preoperative diagnosis of a double jejunal intussusception and jejunal occlusion was based on the findings of small bowel enema and computed tomography. The diagnosis was confirmed at laparotomy.
Key words: Peutz-Jeghers symdrome, jejunal intussusception.

Introduction

The Peutz-Jeghers syndrome first described by Peutz in 1921 (1) in a Dutch family and later by Jeghers in 1949 (2), is an inherited syndrome of hamartomatous gastrointestinal polyps and mucocutaneous pigmentation. Recently the molecular basis of the syndrome was described as a germline mutation in the STK11/LKB1 gene on chromosome 19 p13.3 (3). Several studies have shown an important neoplastic potential for gastrointestinal and extraintestinal tumors. Possible surgical complications are gastrointestinal obstruction, bleeding and perforation. The obstructive ileus can be caused directly by a voluminous polyp or indirectly by means of intussusception (4). We report the case of a 27-year-old woman with an intussusception of the small bowel due to Peutz-Jeghers syndrome.

Case report

A 27-year-old woman was admitted to our hospital with a history of recurrent abdominal pain, abdominal distention and intermittent vomiting lasting 5 months. On physical examination the abdomen showed a mildly tender mass, measuring 5x6 cm located in the left upper quadrant area. Multiple melanin pigmentation of the perioral skin and mouth was also noted (Fig. 1).

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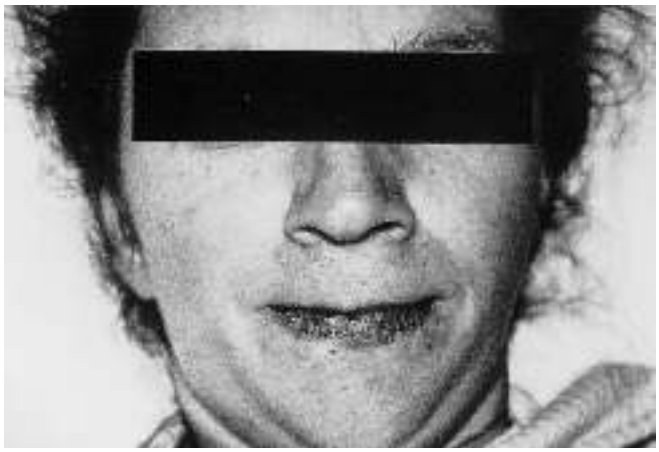

Fig. 1 Multiple melanin pigmentation of the perioral skin and buccal mucosa.

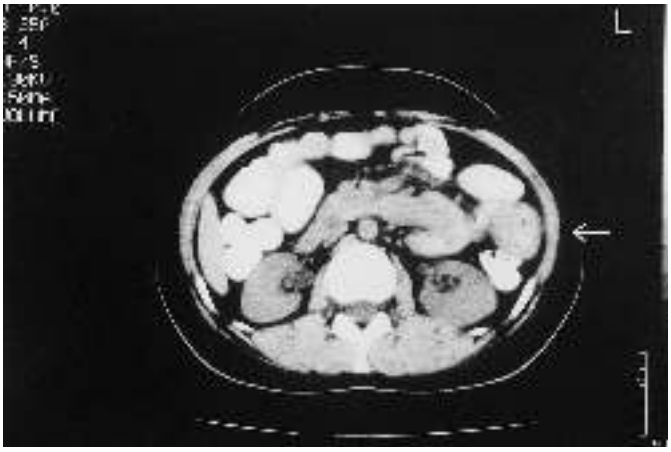

Fig. 2 Abdominal computed tomography scan with enhancement showed a mass occupying the left side of the upper intra abdominal area, possibly corresponding to a jejunal polyp with signs of intestinal intussusception

Endoscopy of the upper and lower gastrointestinal tract revealed multiple polyps in the stomach, duodenum and colon. Endoscopic polypectomy was performed and a total of seven polyps were removed. Histopathological examination of the specimens showed hamartomatous and hyperplastic polyps. Abdominal computed tomography scan with contrast enhancement showed a mass occupying the left side of the upper abdomen, possibly corresponding to a jejunal polyp with signs of intestinal intussusception (Fig. 2). Small bowel enema revealed a huge cauliflower-like polyp in the jejunum with prestenotic dilatation of the intestinal lumen.

Laparotomy was performed. The first 30 cm of the jejunum showed mechanical obstruction due to one large polyp; several other palpable smaller polyps were also found close to that location. Limited (15 cm) small bowel resection and end-to-end anastomosis was performed (Fig. 3). 150 cm distally in the jejunum a double intussusception was confirmed. The double jejuno-jejunal intussusception was successfully managed by operative manipulation, two enterotomies and two polypectomies.

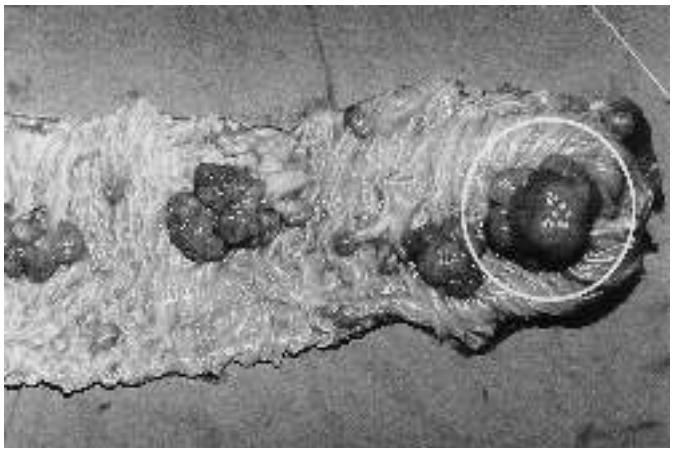

Fig. 3 Small bowel resection (15 cm) specimen of the jejunum with one large and several smaller polyps.

Four other polyps in the remainder small bowel were palpable and all removed through enterotomies. The histological results confirmed the typical hamartomatous polyps without features of malignancy. The postoperative course was uneventful.

Discussion

The Peutz-Jeghers syndrome is an autosomal dominant inherited disease, characterized by hamartomatous gastrointestinal polyposis and mucocutaneous pigmentation. The syndrome is rare; an incidence of 1/60 000 to 1/300 000 newborns has been described in US, with an unknown worldwide distribution (5). The male/female ratio is 1:1. The genetic and molecular basis of the syndrome is a germline mutation in a tumour suppressor gene, the serine-threonine kinase 11(STK11) coding for a protein of the family of serine-threonine kinases, located on chromosome19p13.3 (3). Hamartomas and carcinomas in patients with the Peutz-Jeghers syndrome show loss of heterozygocity at chromosome 19p13.3, indicating inactivation of the wild-type STK11/LKB1 gene (3).

The hamartomatous polyps are located in the jejunum and ileum (90% of the cases), colon (9%) and stomach (24%). 16% of the polyps have also an adenomatous component, most frequently located in the duodenum and jejunum. The incidence of neoplastic change in these polyps is between 3% and 6% at the time of diagnosis (6). There is no relation between the size of the polyps and the neoplastic character (4). Intestinal polyps may give rise to complications such as intestinal occlusion and intussusception (45%), abdominal pain (23%), hemorrhage (14%) and rectal prolapse of a polyp (7%). In the present report, an occlusion was observed in the

entry part of the jejunum because of a large polyp, and a double intussusception in the distal part due to two polyps. Patients with Peutz-Jeghers syndrome show also a higher predisposition for extraintestinal tumours such as pancreas adenocarcinoma, bilateral breast carcinoma and tumours of the urogenital tract (5). The median age of developing cancer is 40 years (42.9±10.2). In a study with a median follow-up of 20 years, 53% of the patients developed multiple cancers such as: breast (54%), colon (39%), pancreas (36%), stomach (29%), small intestine (13%), uterus (9%) and esophagus (0.5%) (7).

Endoscopic polypectomy of any hemorrhagic or large polyp should be performed every 2 years after the initial diagnosis. Surgery or laparoscopy combined with intraoperative enteroscopy is indicated for removal of any symptomatic polyp of the small bowel or polyp larger than 1.5 cm (8, 9). In this way, a number of enterotomies can be avoided, and nonpalpable polyps can be removed. Routine breast and gynecologic screening is recommended as a part of the surveillance.

In conclusion, complications from the Peutz-Jeghers syndrome have to be treated endoscopically at first and, if needed, with a conservative surgical approach. Bowel resections must be kept to an absolute minimum (10).

References

1) Peutz J.L.: *A highly remarkable combination of polyposis of the mucosa of the intestinal tract and of the nasopharynx, together with typical mucocutaneous pigmentations of the skin and mucous membranes (Dutch).* Ned Maandschr Geneesk, 10:134-136 1921.

2) Jeghers H., Mc Kusick V.A., Katz K.H.: *Generalized intestinal polyposis and melanin spots of the oral mucosa, lips and digits: A syndrome of diagnostic significance.* N Engl J Med, 241:993-1006, 1949.

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

4) Lazaridis C.h., Papaziogas B., Atmazidis K., Kalaitzis E., Pavlidis T., Papaziogas T.: *Unusual complications of the Peutz-Jeghers syndrome in two consecutive generations of the same family* (German) Zentralbl Chir, 127:147-150, 2002.

5) Cherki S., Adham M., Bizollon T., Gaudin J.L., Baulieux J.: *Intussusception of the small bowel due to Peutz-Jeghers syndrome: a case report and literature review* (French). Ann Chir, 127:714-717 2002.

6) Kilgus M., Rothlin M.A., Largiader F.: *Intussusception of the small bowel due to Peutz-Jeghers syndrome: a case report.* Hepatogastroenterology, 45:454-455, 1998.

7) Giardiello F.M., Brensinger J.D., Tersmette A.C., Goodman S.N., Petersen G.M., Booker S.V., Cruz-Correa M., Offerhaus J.A.: *Very high risk of cancer in familial Peutz-Jeghers syndrome.* Gastroen terology, 119:1447-1453, 2000.

8) Cunningham J.D., Vine A.J., Karch L., Aisenberg J.: *The role of laparoscopy in the management of intussusception in the Peutz-Jeghers syndrome: case report and review of the literature.* Surg Laparosc Endosc, 8:17-20, 1998.

9) Panos R.G., Opelka F.G., Nogueras J.J.: *Peutz-Jeghers syndrome. A call for intraoperative enteroscopy.* Am Surg, 56:331-333, 1990.

10) Hizawa K., Iida M., Matsumoto T., Kohrogi N., Kinoshita H., Yao T., Fujishima M.: *Cancer in Peutz-Jeghers syndrome.* Cancer 72:2777-2781, 1993.

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