
Double-balloon endoscopy and Peutz-Jeghers syndrome: a new look at an old disease

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Peutz-Jeghers syndrome is a rare disease characterized by mucocutaneous hyperpigmentation and intestinal hamartomatous polyposis. Life-threatening complications include intestinal obstruction and increased risk of gastrointestinal malignancies. While colonoscopy continues to serve as the gold standard for examination of the colon, newer techniques such as capsule endoscopy and double-balloon endoscopy (DBE) are now being applied to both treatment and surveillance of this disease.

Capsule endoscopy serves as a minimally invasive means of locating and characterizing polyps in symptomatic patients but is limited to detection only. DBE allows physicians to visit areas of the small bowel that were previously unreachable by older techniques and to treat lesions that are found. By using DBE to treat polyposis, hemorrhagic ulcers, angiodysplasia, strictures, and cancers, the number of small bowel resections can be decreased in these patients.

Double-balloon endoscopy (DBE) is a new technology available to gastroenterologists that bridges the void between the images captured by capsule endoscopy and the need for endoscopic interventions within the small bowel. Prior to DBE, the small bowel was largely inaccessible to endoscopic interventionalists due to its length and multiple, complex loops; complete examination of the small bowel was virtually impossible. Since 2000, capsule endoscopy has revealed images of small bowel lesions previously described predominantly by radiological imaging and pathology specimens. Through DBE, endoscopic interventions can now be done within the small bowel, including stricture dilation, foreign body retrieval, hemostasis with electrocautery, injection or clipping of bleeding sites, biopsy of lesions or masses, and polypectomy.

In the present case report, DBE allowed a novel therapeutic alternative to surgery in a patient with Peutz-Jeghers syndrome (PJS). PJS, a genetic disease, is characterized by multiple pathognomonic hamartomatous polyps throughout the small bowel. The most common complication in adults with this disease is intussusception of the polyps with resultant bowel obstruction and occasionally ischemia sufficiently severe to require surgical resection of the small bowel.

CASE REPORT

A 41-year-old woman had a clinical history of PJS, not confirmed with genetic testing, which had been complicated by small bowel obstruction secondary to intussusceptions requiring bowel resections in 1992 and 2004. The polyps were histologically diagnostic of PJS. The patient presented to the clinic with complaints of mild, episodic epigastric and left upper abdominal discomfort. The pain was characterized by cramping and abdominal bloating lasting hours to days, unrelieved with medications

and responsive only to bowel rest. At the time of presentation, she denied nausea, vomiting, weight loss, change in bowel habits, and signs of gastrointestinal bleeding. The patient reported that this pain was similar to that caused by her previous episodes of small bowel obstruction. She was otherwise healthy and did not take any prescription or over-the-counter medications. Specifically, she did not take nonsteroidal anti-inflammatory drugs. She had a previous history of perioral pigmentation characteristic of PJS; some lesions had spontaneously faded and some had been removed by cosmetic surgery. Her social history was negative for tobacco and alcohol use. She had no family history of colon cancer, polyps, liver disease, coronary artery disease, diabetes, or other tumors characteristic of PJS.

On physical examination, the patient had normal bowel sounds and mild left upper-quadrant tenderness to deep palpation but no masses, hernias, or organomegaly. No guarding or rebound was exhibited. The patient's skin was normal; specifically, there were no residual perioral discolorations.

During the evaluation of the patient's new abdominal symptoms, her gastroenterologist performed a capsule endoscopy examination, which revealed numerous polyps within the stomach and small bowel. Four of those polyps were partially obstructing the lumen of the small bowel. Subsequently, the patient went to Baylor University Medical Center for DBE, the goal of which was to perform a more complete examination

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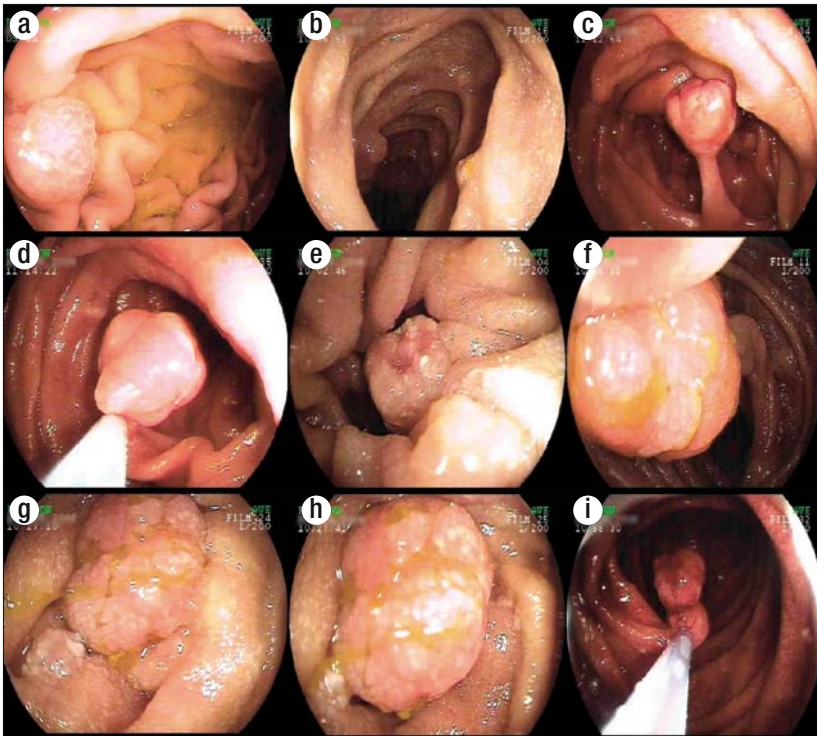


Figure 1. Peutz-Jeghers polyps found during double-balloon endoscopy. (a) Gastric polyp. (b) Sessile polyps in the duodenum. (c) Pedunculated polyp. (d) Polypectomy of an obstructing polyp. (e, f, g, h) Polyps causing nearly complete obstruction. (i) Polypectomy of another polyp.

of the small bowel and to remove any large polyps that could account for the patient's symptoms.

The double-balloon endoscope was introduced through the mouth and advanced past her previous surgical anastomoses to the proximal ileum. The endoscopic examination revealed a normal esophagus, four pedunculated medium-sized polyps in the gastric fundus, and a normal duodenum. The jejunum and ileum demonstrated >20 small hamartomatous polyps and several very large pedunculated polyps measuring up to 20 mm (*Figure 1*). Successful polypectomy and retrieval were accomplished using the inflated overtube balloon to hold the position within the bowel. While the majority of polyps removed endoscopically usually can be suctioned through the 2.8-mm channel of the endoscope, the size of this patient's polyps required that each polyp be snared and brought out through the mouth for retrieval. The endoscope was easily withdrawn and reintroduced into the jejunum and ileum using the inflated overtube balloon to maintain the position in the small bowel.

The polyps were sent for pathological examination and were found to be benign hamartomatous polyps characteristic of PJS.

DISCUSSION

PJS is an autosomal dominant disease that affects an estimated 1 in 50,000 to 1 in 200,000 live births. Patients with PJS have a fourfold increased risk of gastrointestinal and nongastrointestinal cancers, primarily adenocarcinomas, compared with the general population (1). The risk is highest for gastrointestinal cancers occurring in the esophagus, stomach, small intestine, colon, and pancreas (1). Mehenni and colleagues have reported

that the overall risk for gastrointestinal malignancy at age 40, 50, 60, and 70 years is 12%, 24%, 34%, and 63%, respectively (2). In addition to gastrointestinal cancers, extraintestinal organs at increased cancer risk include the lung, breast, uterus, cervix, ovaries, and testes.

Clinically, PJS is characterized by mucocutaneous hyperpigmentation and gastrointestinal polyposis. The pigmented lesions are found in 95% of patients and are caused by pigment-laden macrophages in the dermis. They are evident within the first year of life (often at birth) and increase in size and number over time. Most of the pigmented lesions fade after puberty except for the perioral pigmentation, which may persist throughout life. The lesions are described as macular, blue-gray pigmentations occurring most commonly on the lips and perioral region. Less commonly, they are identified on the hands, feet, buccal mucosa, nose, perianal area, genitals, and, rarely, within the intestines. Malignant transformation of the mucocutaneous lesions is extremely rare (3).

Gastrointestinal hamartomatous polyps begin growing during the first decade of life, with most individuals becoming symptomatic between the ages of 10 and 30 years. A significant number of patients present with intestinal obstruction caused by intussusception or obstruction of the lumen by a polyp. Other presentations include abdominal pain, hematochezia, or prolapse of a rectal polyp. Polyps most frequently occur in the small intestine and colon but can be found anywhere along the gastrointestinal tract. Usually, there will be 1 to 20 polyps per segment of small bowel, and these polyps vary in size, up to 5 cm in diameter (3).

Treatment consists of polypectomy of polyps >1 cm found during endoscopic surveillance. Surgery is reserved for acute obstruction. More recently, capsule endoscopy and DBE have been employed to detect and remove small bowel polyps. In a study comparing capsule endoscopy with bowel barium radiographs, capsule endoscopy identified more polyps and was better tolerated by the patients (4). Capsule endoscopy can also provide the location and size of the polyp. DBE provides a mechanism to treat parts of the small bowel that were inaccessible by older techniques.

DBE was developed in Japan by Yamamoto (5–8) and approved by the Food and Drug Administration for use in the USA in 2004. The double-balloon endoscope consists of a 200-cm endoscope within a 145-cm overtube. Both the endoscope and overtube have inflatable latex balloons at their tips that are controlled by a specially designed air-pump controller that inflates the balloons via a switch (*Figure 2*). DBE is performed under conscious sedation with close monitoring by an anesthesiologist. The endoscope can be inserted orally to examine the small bowel from an antegrade approach, or it can be introduced into the terminal ileum through the colon. In the antegrade approach, the endoscope and overtube are inserted

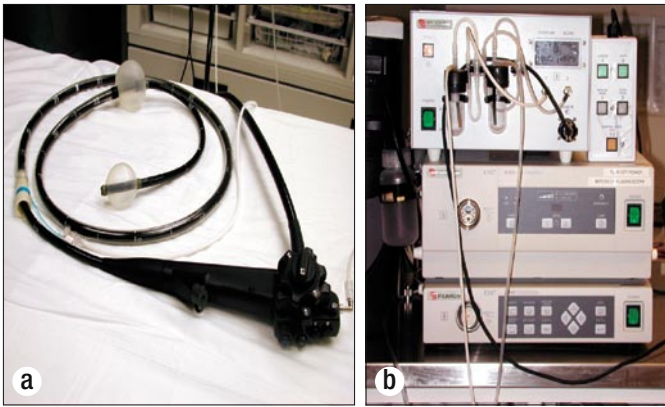


Figure 2. (a) Double-balloon endoscope with a 200-cm scope inside a 145-cm overtube, with inflated latex balloons at each tip. (b) The air-pump controller, which inflates the balloons via a switch.

into the stomach. As the endoscope enters the second portion of the duodenum, the endoscope balloon is inflated. The overtube is advanced to meet the inflated endoscope balloon, and the overtube balloon is also inflated there. With both balloons inflated, the endoscope and overtube are gently pulled back together, thereby shortening and pleating the small intestine over the tube. Next, the endoscope balloon is deflated and the endoscope is advanced further while the inflated overtube balloon holds the position within the small bowel. When the endoscope cannot go any further, the balloon is inflated. The overtube balloon is then deflated, and the overtube is advanced to meet the endoscope. By sequentially inflating and deflating the balloons in this manner and alternately advancing and withdrawing the endoscope, the small bowel is gathered and shortened over the endoscope, allowing for deep insertion into the small bowel (5–8).

DBE is a relatively safe procedure with risks similar to those of colonoscopy. Yamamoto and colleagues reported two cases of complications out of 178 DBE examinations. One complication

was a perforation in a patient with malignant lymphoma of the small intestine recently treated with chemotherapy. The other reported complication was fever and abdominal pain occurring in a patient with Crohn disease. Only one of these complications required surgical intervention (8).

DBE is an excellent innovative technique that provides an alternative to surgery with faster recovery time, decreased length of hospitalization, and decreased cost overall. By utilizing DBE to perform interventions within the small bowel, it is possible to prevent intestinal obstruction, surgical intervention, and loss of the small bowel to a vascular catastrophe. Currently, DBE is available in only 10 cities across the USA. As more facilities adopt DBE, it will have a substantial impact on the way diseases of the small bowel are diagnosed and treated.

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