Small intestinal adenocarcinoma in Peutz-Jeghers syndrome

M V Mehta, Mehul M Porecha, Purvi J Mehta*

Departments of Surgery and *Anesthesiology, M P Shah Medical College, G G Hospital, Jamnagar 361 008

Peutz-Jeghers syndrome (PJS) is characterized by intestinal hamartomatous polyposis (usually affecting the jejunum) and mucocutaneous melanin spots. Though malignant changes are not common, PJS can predispose to carcinoma in the GI tract and elsewhere. We report a 25-year-old man with PJS who developed small intestinal adenocarcinoma and presented with small bowel obstruction due to jejuno-ileal intussusception. [Indian J Gastroenterol 2006;25:38-39]

Peutz-Jeghers syndrome (PJS) is a rare condition, with estimated incidence of 1 in 29,000 to 1 in 8300 live births.1 Though malignant changes are not common, PJS predisposes to carcinoma of the gastrointestinal tract and other organs. Tumors of the small intestine are remarkably rare.

A 25-year-old man presented with bilious vomiting and abdominal distension since 2 days and absolute con-
stipation since 1 day. On examination there was tachycardia, generalized abdominal distention, guarding and rigidity, and bowel sounds were absent. Per rectal examination was normal. The oral cavity showed melanin spots over the buccal mucosa and lips. Erect abdominal radiograph was suggestive of small bowel obstruction.

Emergency abdominal exploration revealed that the obstruction was due to jejuno-ileal intussusception. On reduction, about 45 cm of jejunum was found to be gangrenous. The invagination was caused by a 2-cm polyp in the jejunum (Fig). Six other polyps varying in size from 0.5 cm to 1.5 cm were found in the small intestine. Resection of the gangrenous intestine along with the portion containing the polyps was done and primary jejuno-ileal anastomosis was performed. The postoperative course was uneventful.

On histology, the 2-cm polyp that had caused intususception and other polyps of size 1.5 cm showed changes of adenocarcinoma; rest of the polyps showed features of benign disease.

PJS is an autosomal dominant syndrome characterized by mucocutaneous melanin spots with intestinal polyposis. Mutation in the serine / threonine kinase gene STK, also known as LKB1, located on chromosome 19p, has been identified in more than 50% of patients with PJS. The life-time risk of mucocutaneous lesions approaches 100%, and the risk of polyps in the small intestine (particularly jejunum) and colon is about 50% and 25%, respectively.

PJS predisposes to malignancy of the gastrointestinal tract, pancreas, breast, lung and reproductive organs.\(^2,3\) The increased incidence of malignancy has been attributed to concurrent presence of adenomatous polyps; a hamartoma-carcinoma sequence has also been considered.\(^4\)

In a series of 34 patients with PJS followed up for a median of 20 years, some form of cancer developed in 53% by average 39.4 years of age.\(^1\) The mean interval from initial diagnosis of PJS to diagnosis of cancer was 19.8 years. In another series,\(^3\) among 72 patients with PJS the relative risk of dying from a gastrointestinal cancer was 13-fold greater than that in the general population and relative risk of dying of any other cancer was 9-fold greater, with 48% chances of dying of cancer by the age of 57 years.

Current recommendations advocate prophylactic endoscopic removal of all polyps.\(^2\) Recent guidelines support complete colorectal surveillance with either colonoscopy or flexible sigmoidoscopy with barium enema at 18 years of age and every 3 years thereafter.\(^5\) Upper gastrointestinal endoscopic surveillance is recommended every 1-2 years from age of 25 years.\(^5\) Others have advocated routine small bowel imaging surveillance every 2 years, with laparotomy and resection for polyps greater than 1.5 cm in diameter.\(^2\)

In our patient, the age of detection of small intestinal malignancy was lower than that reported in literature. Surveillance for gastrointestinal, pancreatic, breast and ovarian cancer is appropriate in these patients.

References

Correspondence to: Dr. Porecha, “Ashutosh”, Opp. Ahir Boarding, Amber Cinema Road, Jamnagar 361 008. E-mail: m e h u l m p o r e c h a @ r e d i f f m a i l . c o m , mehulmporecha@hotmail.com

Received March 21, 2005. Accepted June 24, 2005

Successful closure of spontaneous esophageal perforation (Boerhaave’s syndrome) by endoscopic clipping

Parupudi V J Sriram, Guduru V Rao,* D Nageshwar Reddy

Departments of Gastroenterology and *Gastrointestinal Surgery, Asian Institute of Gastroenterology, Hyderabad

Endoscopic clips have been used mainly for control