This photograph is from an upper gastrointestinal endoscopy on a 15-year-old male. He has a history of a total colectomy and is being evaluated for iron deficiency anemia. He denies abdominal pain, weight loss, and melena. He notes occasional bright red blood on the toilet paper but denies hematochezia.

Test your diagnostic skills with the following endoscopic quiz.

1) The photograph is a view of the descending duodenum and demonstrates:
   a) Multiple ulcers
   b) An ampullary mass
   c) A duodenal diverticulum
   d) Multiple polyps
   e) Villous atrophy

2) The most likely diagnosis is:
   a) Zollinger-Ellison syndrome
   b) Familial adenomatous polyposis
   c) Scleroderma
   d) Juvenile polyposis
   e) Celiac disease

Answers to 'Diagnostic Dilemma: GI Disease'
1. The correct answer is (d), multiple polyps. There are several polyps ranging from approximately 4 to 7 mm in diameter. The photograph does not reveal ulcerations, a mass, diverticulum, or evidence of atrophic mucosa.
2. The correct answer is (b), familial adenomatous polyposis (FAP), an inherited autosomal dominant
disorder associated with the development of colon cancer. It is caused by a germline mutation in the adenomatous polyposis coli (APC) gene located on chromosome 5q21-22, and is characterized by the development of more than 100 adenomas in the colorectum.

The lifetime risk of colorectal carcinoma is virtually 100% if patients are not treated by colectomy. The primary cause of death in this syndrome is colorectal cancer, which develops generally by the 5th decade of life.[1]

As a result of the decline in colon cancer deaths in patients with FAP, deaths due to the development of duodenal adenocarcinoma are on the rise. The duodenum is the second most commonly affected site of polyp development in FAP. Polyps can be found in 30% to 70% of patients with FAP, with the lifetime risk of these lesions approaching 100%.

Duodenal adenomas tend to occur at a later age and have a lower potential for malignant change, compared with colon polyps. However, patients with FAP have a 100 to 330 times higher risk of developing duodenal cancer, compared with the general population.[1]

It is estimated that about 5% to 10% of patients with duodenal polyps will eventually develop duodenal adenocarcinoma.[2-4] and it is the second leading cause of death in FAP patients.[1,2]

The most useful system for rating the severity of duodenal polyposis was developed by Spigelman and colleagues. This system describes five stages (0 to IV) of duodenal polyps, with points given based on the number of polyps, size, histology, and severity of dysplasia. Studies have shown that most patients with duodenal polyposis will slowly progress through the stages, with almost all cases of adenocarcinoma occurring in patients with stage IV disease.[3]

This Spigelman system is used in guiding surveillance and management strategies. It has also been found to correlate with the risk of duodenal malignancy, with stages II, III, and IV associated with a 2.3%, 2.4%, and 36% risk of duodenal cancer, respectively.[1]

Prevention strategies

The increased relative risk of duodenal carcinoma in FAP patients and the poor outcomes associated with the treatment of advanced duodenal cancer have led to the development of prevention strategies via surveillance.

Surveillance of FAP patients should begin at about 21 years of age and should be performed using both an end-viewing and a side-viewing upper endoscope. If no polyposis is evident, follow-up should be performed at an interval of 3 to 5 years. If polyposis develops (stage I or II disease), screening is performed at an interval of 1 to 3 years.

Patients with denser polyposis or larger adenomas (stage III or IV disease) should undergo examination every 6 to 12 months because of their increased risk of duodenal adenocarcinoma.[3]

Prophylactic surgery should be strongly considered for stage IV disease.

Adenomatous polyps can occur in the antrum, and a small fraction of fundic gland polyps contain adenomatous foci that may progress. Representative biopsy samples of fundic gland polyps and removal of larger or suspicious-appearing lesions through endoscopy are recommended.

Although supporting literature is lacking, push enteroscopy, enteroclysis or CT enterography, and wireless capsule endoscopy should also be considered for surveillance of jejunal and ileal neoplasms at the same intervals as for duodenal polyps.[3]

Treatment

Treatment of duodenal polyposis is difficult. Endoscopic snare polypectomy and ablation of as many of the most significant polyps as possible should be performed, although duodenal adenomas tend to recur within a year.

The more radical Whipple procedure and pylorus-preserving duodenectomy are most definitive for reducing the risk of adenocarcinoma. However, these procedures are associated with significant morbidity and mortality, including the risk of inducing desmoid tumor formation in about 10% of FAP patients. A small but real risk of jejunal and ileal carcinoma persists in these patients.

As operative or endoscopic measures fail to control duodenal adenomas in FAP patients, interest has focused on drug therapies. Although the evidence is limited, nonsteroidal anti-inflammatory drugs (NSAIDs)[je, sulindac, a nonselective cyclooxygenase (COX) inhibitor, or celecoxib (Celebrex), a COX-2 selective inhibitor[may be of benefit after the development of duodenal polyposis by inducing the regression or stabilization of the polyposis.[5-7]

References:


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