Upper Gastrointestinal Neoplasia in Familial Polyposis

ROBERT C. KURTZ, MD, STEPHEN S. STERNBERG, MD, HELEN H. MILLER, MSW, and JEROME J. DECOSE, MD

Upper gastrointestinal (UGI) endoscopy was performed in 41 asymptomatic American patients with familial polyposis to assess the prevalence of gastric and duodenal polyps and to characterize their pathological features. Eighteen patients (44%) had UGI endoscopic abnormalities. Six patients had both gastric and duodenal lesions. Eight patients had only gastric polyps, and four had duodenal polyps only. The presence of other extracolonic expressions of polyposis had a suggestive but statistically insignificant correlation with UGI polyps. Patients with familial polyposis and duodenal adenomatous polyps are at high risk for the development of periampullary cancer; screening and identification of these individuals is recommended.

KEY WORDS: UGI polyps; familial polyposis.

Familial polyposis is a hereditary premalignant disorder which leads almost invariably to large bowel cancer if left untreated (1). As awareness of colonic polyposis has increased and prophylactic removal of the large bowel has been performed more widely, other inherited, life-threatening expressions, particularly upper gastrointestinal cancer and desmoid tumors have received increasing attention (2, 3).

Benign neoplasia in the upper gastrointestinal tract of polyposis patients has long been recognized (4-6), but only recently, stimulated by Japanese investigators (7-9), have endoscopic surveys of the prevalence of gastric and duodenal polyps been reported (10-14). Taken together, these reports identify a frequency of gastric polyps ranging from 39% to 100% and of duodenal polyps ranging from 46% to 93%. Most gastric polyps have been found to be hyperplastic, whereas most duodenal polyps have been adenomatous. Adenomatous changes have been also identified frequently in biopsies of the ampulla of Vater. Other reports have cited the presence of polyps throughout the small intestine (15-17).

In 1935, Cabot first reported periampullary cancer in a polyposis patient (18). Subsequent investigators have added further evidence of this association; polyps and cancer have also been observed more distally in the duodenum, the jejunum, and the ileum (16, 19, 20), as well as in bile duct and gallbladder epithelium (21, 22). Although gastric cancer has been linked with polyposis, there has been only one instance of this association reported from the United States (23).

Upper gastrointestinal neoplasia has not generally been regarded as a component of Gardner's
syndrome. In 1953, Gardner and Richards (24) first defined the syndrome of familial polyposis with extracolonic expressions of epidermoid cysts, osteomas, and fibromas; later (25) they added dental abnormalities and desmoid tumors. As polyposis patients have been more intensively studied, these and other extracolonic associations have been observed to occur frequently, either in the proband or in a relative, and the utility of this eponymic distinction among polyposis cases has been questioned (2, 3, 10, 12).

We examined a group of 41 asymptomatic polyposis patients to assess the prevalence of gastric and duodenal polyps and to address their features, significance, and management. In this study, the term “polyposis” is used to designate the disease of all patients in our population.

MATERIALS AND METHODS

Upper gastrointestinal endoscopy was performed in 41 asymptomatic polyposis patients of the total study group of 62 polyposis patients entered in a chemoprevention trial. All 62 patients were offered upper gastrointestinal endoscopy but only 41 accepted. All 62 study patients had had a total abdominal colectomy and ileorectal anastomosis. Of the 41 patients who underwent endoscopy, 26 had them performed at Memorial Sloan-Kettering Cancer Center (MSKCC), 23 by one endoscopist, and the remaining 15 procedures were performed by endoscopists geographically convenient to the patient’s home. At MSKCC, the procedure was performed with Olympus GIFXQ and GIFQ endoscopes. Informed consent was obtained from each patient.

One physician evaluated each patient for the presence and type of extracolonic expressions by careful physical examination. These findings were correlated with the observations made at upper gastrointestinal endoscopy. Endoscopic biopsies of representative gastric and duodenal polyps were obtained and reviewed by one pathologist.

RESULTS

Endoscopic Surveillance. Upper gastrointestinal polyps were found in 18 of 41 asymptomatic polyposis patients (44%). Among the 18 affected patients, the gastric mucosa alone was involved in eight (20% of total), the duodenal mucosa alone in four (10%), and the mucosa of both organs was involved in six (15%). The detection rate of polyps was the same whether the endoscopy was performed at MSKCC (12 of 26, 46%) or elsewhere (6 of 15, 40%).

Ten of 20 men (50%) and eight of 21 women (38%) had polyps. The mean age of the 10 men without polyps was 36.0 years. The mean age of the eight women with UGI polyps was 32.5 years, whereas that of the 13 women without polyps was 29.9 years. Neither patients’ sex nor age were significant variables. The distribution by age and sex of the trial patients who declined endoscopy paralleled those who had endoscopic evaluation.

Other extracolonic expressions of polyposis were found in 11 of 18 patients with UGI polyps (61%) and in nine of 23 without UGI polyps (39%) (Table 1). These differences were suggestive but not statistically significant (P = 0.14, Fisher exact test). Hence, 20 of the 41 patients (49%) in this endoscopic study had other extracolonic expressions; among all 62 patients entered in the chemoprevention trial, extracolonic expressions were found in 34 (55%).

Moreover, the type of extracolonic expressions did not differ between those patients with and those without UGI polyps. Among the 11 with UGI polyps, extracolonic expressions included: sebaceous cysts alone in four, sebaceous cysts and lipomas or fibromas in four, sebaceous cysts and papillary thyroid cancer in two, and osteomas in one. Among the nine without UGI polyps, extracolonic expressions included: sebaceous cysts alone in three, sebaceous cysts and lipomas or fibromas in three, sebaceous cysts and osteomas in one, lipomas only in one, and an intraperitoneal desmoid tumor and fibromatosis in one.

Endoscopic Findings. Gastric polyps were concentrated in the fundus and body of the stomach. Usually these polyps appeared as sessile nodules or mucosal excrescences, although a rare polyp had a narrow stalk. Polyps were very small, usually 2–3 mm in diameter, with several as large as 5 mm. They were diffuse or in clusters and were always multiple, ranging in number from two to more than 100, with the higher numbers being most common. In only two instances were gastric polyps confined...