A normal appearing duodenal papilla in familial adenomatous polyposis needs to be biopsied: A case report

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FAMILIAL ADENOMATOUS POLYPOSIS (FAP) is a rare hereditary disease transmitted in an autosomal dominant pattern. The gene recently has been isolated on chromosome 5q (1). This genetic defect predisposes affected patients to develop neoplastic polyps involving the entire gastrointestinal tract (2). Innumerable colorectal adenomas occur at a young age and there is inevitable progression to adenocarcinoma, usually by age 40 years, unless the colon is removed (3); this has led to the widely accepted treatment of surgical resection of the entire colon once the disease has been identified.

The association between FAP and upper gastrointestinal neoplasms was first described 100 years ago (4). It was not until the 1970s that Japanese investigators (5,6) pointed out the high incidence of gastrointestinal tumours in patients with FAP. These studies have been confirmed over the past two decades in western nations, resulting in recommendations to screen patients with FAP endoscopically for gastrointestinal neoplasms (7). Periampullary tumours also may be quite common in families with the recently described hereditary flat adenoma syndrome (8).

We report an asymptomatic patient with FAP who was found on biopsy to have an adenocarcinoma of the papilla of Vater that endoscopically was of normal appearance.
CASE PRESENTATION

A 50-year-old man with known FAP was referred for endoscopic screening. The patient’s father, paternal grandfather and a paternal aunt had FAP and colon cancer in their 30s. A cousin had a total colectomy when FAP was found on screening sigmoidoscopy. The patient had undergone a subtotal colectomy and ileorectal anastomosis five years earlier for asymptomatic polyposis.

The patient’s rectum was followed over the next five years by semi-annual sigmoidoscopies, and several adenomas were removed by polypectomy during this period. The patient was completely asymptomatic and had normal bowel movements. The physical examination apart from a midline abdominal scar was normal. An ophthalmologist demonstrated retinal hypertrophic pigmentation in the left eye.

Complete blood count and liver enzymes were normal. Upper endoscopy revealed multiple 2 to 5 mm polyps in the fundus and body of the stomach. The antrum pylorus and duodenum were normal. Biopsies and polypectomy confirmed fundic gland polyps.

With the use of a videoduodenoscope, two minipolyps were removed from the second portion of the duodenum by biopsy; these consisted of lymphoid hyperplasia histologically. The papilla was prominent and covered by normal looking mucosa (Figure 1). It had two separate openings. Random biopsies were obtained from both papillary orifices and the body of the papilla, revealing the presence of moderately differentiated adenocarcinoma (Figure 2).

Subsequently, the patient underwent endoscopic retrograde cholangiopancreatography (ERCP) (Figure 3). Contrast injection of the proximal papillary orifice yielded a normal cholangiogram whereas cannulation of the distal orifice produced a normal pancreaticogram. A computed tomographic (CT) scan of the abdomen showed a mass within the mesentery in the left upper quadrant contiguous with several small bowel loops in keeping with a desmoid tumour. The pancreas looked normal.

The patient initially underwent local excision of the ampulla but frozen section demonstrated adenocarcinoma with invasion of the duodenal wall, and a complete Whipple procedure was carried out.

At one year follow-up the patient was doing well without any evidence of tumour recurrence. Shortly thereafter, however, an ultrasound demonstrated multiple hepatic filling defects and a liver biopsy confirmed the diagnosis of metastatic adenocarcinoma.

DISCUSSION

As prophylactic colectomy extends the lives of polyposis patients extracolonic tumours are gaining increasing clinical significance. Gastric polyps occur in more than 50% of affected individuals and usually are fundic gland polyps histologically with no premalignant potential (9-11). Although Japanese studies (12,13) report a similar incidence of gastric adenomas these neoplasms are found in fewer than 8% of FAP cases described in western nations (11,14).

The duodenum is by far the most common site of extracolonic tumours in patients with FAP or its phenotypic variant, Gardner’s syndrome. The prevalence of duodenal adenomas in FAP exceeded 90% in a recent series when side-viewing videendoscopy was combined with directed and randomly taken duodenal biopsies (11).

The tumours are predominately distributed in the second and third parts of the duodenum with prominent periampullary clustering (15-17) although more distal lesions have been reported as well (18).

There is a high concentration of...
patients have a genetically determined mucosal growth abnormality rendering them more susceptible to tumour promoters or whether abnormal cell proliferation arises because of altered concentrations of promoters such as bile acids. Both situations might coexist resulting in the formation of adenomas (19).

There is strong evidence that most, if not all, ampullary and duodenal carcinomas develop in pre-existing adenomas following an adenoma-carcinoma sequence similar to that accepted for colorectal cancer (20-22).

The patient described in this report had an endoscopically normal appearing papilla and there was no symptomatic, biochemical or radiographic evidence on CT scan or ERCP for a malignant ampullary tumour.

Periampullary carcinomas represent the leading cause of cancer-related deaths in FAP patients after colectomy (2,16). A recent study (23) estimated that the relative risk of duodenal and periampullary adenocarcinoma is greatly increased in FAP patients, with absolute risk of one case per 1698 person-years. Screening FAP patients for upper gastrointestinal neoplasms is widely recommended (7,17,23,24). Although the optimal frequency of screening has not yet been established, several authors (7,16,25) have outlined their protocols recommending surveillance endoscopy and biopsies every three to five years with the initial study carried out at colectomy. The examinations should be carried out using both end- and side-viewing instruments (7). This case report emphasizes the need for biopsies of the major duodenal papilla even in the absence of a visible abnormality.

REFERENCES