Familial Polyposis in Children

DONALD A. PECK, M.D., K. S. WATANABE, M.D., H. W. TRUEBLOOD, M.D.

From the Department of Surgery, Stanford University, Santa Clara Valley Medical Center, San Jose, California

Familial polyposis of the colon may occur in the early years of life. The disease can be diagnosed via routine examination of the polyposis family or as a result of the investigation of diarrhea and/or the passage of blood per rectum. Management in childhood should include prompt colectomy, since a small but significant number of cases of carcinoma complicating familial polyposis in children have been reported.

Incidence

Familial polyposis is hereditary, transmitted via a non-sex-linked mendelian dominant gene. If the affected parent is heterozygous, half of the children will inherit the disease. Reed and Neel estimate the minimum incidence of individuals with the gene for polyposis at birth to be one in 8,300. There is considerable variation in the ages of appearance of the disease in affected members of polyposis families.

The experience recorded by Dukes in Britain suggests that patients with polyposis appear normal at birth, and often throughout childhood. Dukes reported that symptomatic polyposis prior to the second decade of life is rare. McKenney reviewed the ages at which polyposis was diagnosed in 21 affected members of three polyposis families. Eleven of the 21 polyposis patients were diagnosed prior to the age of 16 years. Seventy-five cases of multiple polyposis in patients less than 13 years old were reported in Abramson's review.

LeFevre and Jacques recorded the case of a 4-month-old infant with symptomatic prolapsing polyposis which led to death from gangrene and intussusception. Ravitch reported a 10-month-old infant with familial polyposis whose life ended in a similar fashion. These cases suggest that colonic polyposis may be present during intrauterine life.

Report of Three Cases

Patient 1. A 9-year-old asymptomatic boy was diagnosed via proctoscopy in December 1969. The rectal mucosa had a cobblestone appearance, and many prominent polyps were visualized. Biopsy confirmed the presence of adenomatous polyps.
Fro. 2. Cecum and terminal ileum. Filling defects in the terminal ileum are the result of submucosal lymphoid hyperplasia.

Proctosigmoidoscopy and air-contrast barium-enema studies (Fig. 1) confirmed the diagnosis of multiple polyposis of the colon. The appearance of the terminal ileum suggested lymphoid hyperplasia (Fig. 2).

The patient's mother had undergone colectomy for familial polyposis at the age of 31 years. His maternal grandmother had had polyposis and had died of metastatic adenocarcinoma of the sigmoid at the age of 37 years.

In June 1970, when he was 10 years old, the patient underwent total colectomy and ileoprostostomy with preservation of the ileocecal valve (Fig. 3). Multiple polyps identified in the terminal 12 inches of ileum were indistinguishable in appearance from adenomatous polyps. The final pathology report indicated multiple adenomatous polyps of the colon (Fig. 4). A biopsy of the terminal ileum reflected submucosal lymphoid hyperplasia.

The rectal polyps were not fulgurated prior to surgery, nor were they treated in the postoperative period. Quarterly proctosigmoidoscopic examinations have shown spontaneous remission of all residual polypoid disease. Growth and development have continued normally. The patient has an average of two stools per day.

Patient 2. The 7-year-old younger brother of Patient 1 was examined in January 1970 and the diagnosis established via proctosigmoidoscopy, biopsy and air-contrast barium-enema studies. In June 1970, he underwent colectomy and ileoprostostomy with preservation of the ileocecal valve. Air-contrast barium-enema studies revealed lymphoid hyperplasia in the terminal ileum.

Fig. 3. Preservation of the ileocecal valve. A, line of incision in the cecum and ileocolic mesentery preserving the ileocolic artery. B, prepared terminal ileum, ileocecal valve and cuff of cecum. C, cecal cuff anastomosed to upper rectum.