Malignant Potential of Juvenile Polyposis Coli
Report of a Case and Review of the Literature

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Juvenile polyps of the colon and rectum traditionally have been viewed as being benign inflammatory or hamartomatous lesions without potential for malignant change. The authors report a case of adenocarcinoma developing in a patient with sporadic juvenile polyposis. Juvenile polyposis was diagnosed in the patient at age 4 years. He underwent subtotal colectomy at age 6 years. At age 12, he underwent a proctectomy and a Swenson pull-through because of adenomatous changes in the rectal stump. At age 19 surveillance endoscopy revealed invasive cancer in a juvenile polyp. [Key words: Juvenile polyposis; Nonfamilial; Adenomatous hyperplasia; Adenocarcinoma rectum]

The juvenile polyp is the most common form of tumor of the colon in children. These lesions occur in 1 to 2 percent of children, usually within the first decade of life, but also may be seen in older children as well as adults at any age. They usually occur either as a solitary polyp or as two or three isolated polyps. Less frequently they take the form of a generalized polyposis, which is often familial (probably autosomal dominant), but may also occur as a sporadic event. Isolated juvenile polyps often present with bleeding and prolapse but follow an innocuous course with autoamputation a common endpoint. Operative intervention is limited to the rare complications such as hemorrhage, intussusception, intractable diarrhea, or failure to thrive. Generalized juvenile polyposis causes more severe diarrhea, bleeding with profound anemia, and hypoproteinemia. These polyps tend to persist into adult life, with adenomatous changes occurring commonly. In this group of patients, family members may be at risk for development of gastrointestinal tract neoplasms.

The isolated juvenile polyp is a benign hamartomatous or inflammatory lesion with negligible potential for malignancy. Foci of neoplastic (adenomatous) transformation have been described more often in patients with generalized juvenile polyposis, but only rarely have these adenomatous polyps been reported to develop into frank malignancy within the colon and rectum. We present an unusual case of a patient with nonfamilial generalized juvenile polyposis who developed invasive carcinoma in a background of adenomatous change in juvenile polyps of the rectum. The purpose of this report is to draw attention to the established but underappreciated association of juvenile polyps and adenocarcinoma.

Report of a Case

A previously healthy four-year-old boy presented with a one-week history of rectal bleeding. Barium enema (Fig. 1), endoscopy, and biopsies revealed numerous juvenile (retention) polyps in the
colon and rectum (Fig. 2a). There was no family history of polyposis syndrome or gastrointestinal malignancy. Initially the polyps were removed endoscopically, until, at age 6, a random biopsy of a colonic polyp revealed adenomatous changes within a juvenile polyp (Fig. 2b). At that time, the patient was noted to have a hemoglobin level of 7.8 and an albumin level of 2.5, with marked osteoarthropathy of his fingers and toes (Fig. 3). Subsequently, he underwent a subtotal colectomy and ileosigmoidostomy with marked improvement in his systemic findings (Fig. 4). Yearly evaluation continued until age 12, when again a biopsy showed adenomatous changes. Consequently he underwent a Swenson pull-through procedure leaving a 2-cm cuff of intact rectal mucosa when endorectal excision of the polyp-laden mucosa in this area became technically impossible. At age 17 he was noted to have an adenomatous 10 x 12 cm rectal polyp, which was prolapsed, friable, and bleeding. He was taken to the operating room where this polyp as well as an additional 50 gm of polyp tissue were removed. The large polyp was found to be a tubulovillous adenoma.

Surveillance proctoscopy at three-month intervals continued in view of the family's request to delay completion proctectomy. At age 19 years a rectal biopsy showed dysplasia with a focus of intramucosal adenocarcinoma within a juvenile polyp (Fig. 5a, b). Exploration of the abdomen revealed dense adhesions and a number of enlarged lymph nodes. The entire rectum as well as 12 cm of terminal ileum were removed and an end ileostomy created. The surgical specimen was found to contain poorly differentiated adenocarcinoma of signet-ring cell type in the rectum and extending into the terminal ileum 3 cm proximal to the previous ileorectal anastomosis with extensive involvement of extramural soft tissue (Fig. 6a, b). The patient was begun on 5-fluorouracil and leucovorin for presumed locally advanced disease. Postoperatively a computed tomographic (CT) scan of the chest, abdomen, and pelvis revealed no evidence of metastatic disease, but arteriovenous malformations (AVMs) were noted in the left lower lung field (Fig. 7). Ileoscopy revealed numerous polyps, less than 1.0 cm, 20 cm proximal to the ileostomy.

Two months after proctectomy, a "second look" abdominal procedure revealed a solitary 4 x 5 cm metastatic mass embedded in the mesentery of the pelvic inlet, which was grossly resected. This proved to be a lymph node metastasis with perinodal soft tissue extension. Before discharge, the pulmonary AVMs were obliterated by balloon embolization. Sixteen months after surgery the