Attenuated Familial Adenomatous Polyposis Associated with Advanced Rectal Cancer in a 16-Year-Old Boy: Report of a Case

SHIGETOSHI MATSUO1, SUSUMU EGUCHI1, TAKASHI AZUMA1, MASAAKI HIDAKA1, SATOSHI YAMAGUCHI1, TOMAYOSHI HAYASHI2, NORIHiro KOHARA3, and TAKASHI KANEMATSU4

Departments of 1Surgery and 2Pathology, Nagasaki Prefectural Shimabara Onsen Hospital, 7895 Shimokawajiri, Shimabara, Nagasaki 855-0861, Japan
3Department of Surgery, Nagasaki Municipal Hospital, 6-39 Shinchi, Nagasaki 850-8555, Japan
4Department of Surgery II, Nagasaki University School of Medicine, 1-7-1 Sakamoto, Nagasaki 852-8501, Japan

Abstract We herein present a case of attenuated familial adenomatous polyposis (AFAP) with advanced rectal cancer in a 16-year-old boy. His mother and younger brother both had subcutaneous soft tissue tumors in the back and sparse-type colorectal polyposis. His mother also had dental anomalies and gastric fundic gland polyposis. The patient was admitted to our hospital for investigation of bloody stools. Barium enema and colonofiberscopy revealed advanced rectal cancer and sparse (<50) colorectal polyps. He also had dental anomalies, a subcutaneous soft tissue tumor in the back, and gastric fundic gland polyposis as extracolonic manifestations. A total proctocolectomy and ileoanal anastomosis were performed, and histological examination of the resected specimens confirmed moderately differentiated adenocarcinomas of the rectum with metastases to the regional lymph nodes. The other colorectal polyps were tubular adenomas with no evidence of malignancy. Germline mutations in the APC gene were observed in codons 486, 545, 1493, and 1556. This case serves to demonstrate that a total proctocolectomy with ileoanal anastomosis should be the procedure of choice for young patients found to have advanced rectal cancer associated with FAP.

Key words Familial · Adenomatous · Polyposis

Introduction

Familial adenomatous polyposis (FAP) is an autosomal dominant inherited disease mainly characterized by colorectal adenomatous polyposis with a 100% chance of becoming malignant in the future.1–3 Attenuated FAP (AFAP), which is characterized by fewer colorectal adenomas (1–100) mainly distributed in the proximal colon, with a high incidence of gastric fundic gland polyposis and duodenal adenomas, has recently been recognized.4–6 In treating FAP patients, surgeons have tried to determine the ideal time to perform prophylactic colectomy and to decide whether the rectum should be preserved or not. They have also considered the importance of making a presymptomatic molecular diagnosis. This report describes the case of a 16-year-old boy with AFAP and advanced rectal cancer, and reviews the literature on this subject.

Case Report

A 16-year-old boy was admitted to Nagasaki Prefectural Shimabara Onsen Hospital for investigation of bloody stools. His mother had sparse-type colorectal polyposis which had been previously diagnosed as tubular or tubulovillous adenoma from polypectomy specimens, gastric fundic gland polyposis, dental anomalies, and a subcutaneous soft tissue tumor in the back. His younger brother was also known to have sparse-type colorectal polyposis, histologically proven to be tubular adenoma, and a subcutaneous soft tissue tumor in the back. His grandfather had died of gastric cancer in his forties. On admission, complete blood counts and blood chemistry values were within normal limits. The serum level of carcinoembryonic antigen was 4.9 ng/ml (normal range: <5.0 ng/ml). Rectal examination revealed a hard tumor. An ophthalmological examination was normal and a mandibular pantomography revealed the presence of milk teeth and burying teeth. Computed tomography demonstrated thickening of the rectal wall, swollen regional lymph nodes, and a subcutaneous soft tissue tumor in the back. No evidence of tumor was found in the brain or thyroid gland. Gastrofiberscopy revealed fundic gland polyposis. The ampulla of Vater...
was normal and no small bowel polyps were found. Barium enema and colonofiberscopy revealed the presence of advanced rectal cancer with a central ulceration, and fewer than 50 polyps, the majority of which measured about 5 mm at the greatest diameter, were found in the colorectum (Fig. 1). Histological examination of biopsied specimens from the rectal tumor revealed findings of adenocarcinomas, and the other polyps were diagnosed as tubular adenomas. On the basis of these findings, the patient was diagnosed to have AFAP with advanced rectal cancer. On March 23, 2000, a total proctocolectomy with dissection of the regional lymph nodes was performed, and after the construction of a linear stapled J pouch, a handsewn ileoanal anastomosis (IAA) was also carried out (Fig. 2). No evidence of liver metastases or peritoneal disseminations was noted. A temporary diverting ileostomy was performed as an optional procedure. Histological examination of the resected specimens showed moderately differentiated adenocarcinomas and invasion to the adjacent perirectal adipose tissues. A small amount of adenomatous component was partly intermingled with carcinomatous components (Fig. 3). These findings had resulted from an adenoma-carcinoma sequence. Regional lymph node metastases were also seen. A total of 16 visible adenomatous polyps without malignancy were found in other sites in the colorectum. The patient had an uneventful postoperative course and did not suffer any complications.

Under informed consent, sequence analysis of the APC gene was performed as follows: first, total RNA was extracted from lymphocytes; next, the entire coding region of the APC gene was amplified using the polymerase chain reaction (PCR); and finally, the PCR products were analyzed by an ABI 310 Genetic Analyzer. Germline mutations in the APC gene of this patient were observed in codons 486 (polymorphism TAC to TAT), 545 (polymorphism GCA to GCG),