Journal of Pediatric Surgery (2010) **45**, 525–532

ELSEVIER

Journal of Pediatric Surgery

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Evaluation of 28 years of surgical treatment of children and young adults with familial adenomatous polyposis

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Received 12 September 2008; revised 5 June 2009; accepted 12 June 2009

Key words:
Familial adenomatous polyposis coli;
Pediatric patients;
Children;
Teenagers;
Ileal pouch-anal anastomosis;
Ileorectal anastomosis

Abstract

Background: In this retrospective study, 28 years of surgical treatment of children and young adults with familial adenomatous polyposis (FAP) was analyzed.

Methods: Forty-three patients were operated on before the age of 26 years. Endoscopic aspects, operative data, and complications were analyzed, and the resection specimens were reevaluated. Functional outcome was assessed by telephone questionnaire.

Results: Primary ileorectal anastomosis (IRA) was performed in 34 patients with a mean age of 16 years (range, 7-25 years). Primary ileal-pouch anal anastomosis (IPAA) was performed in 9 patients at a mean age of 19 years (range, 15-24 years). Secondary excision of the rectum was performed in 7 patients. Overall, rectal carcinoma was present in 4 patients, at the age of 35, 36, 37, and 38 years. Two patients, aged 39 and 40 years, died because of invasive carcinoma with distant metastasis. The functional outcome and postoperative complications after both procedures were similar to those described in literature for children with FAP. Most patients did not experience alterations in lifestyle, and there was no urinary incontinence.

Conclusions: In this retrospective study, both IRA and IPAA showed to be feasible techniques in young patients with FAP. A prospective study with a sufficient follow-up is needed to compare both techniques in this specific group of patients.

Familial adenomatous polyposis is caused by mutations in the adenomatous polyposis coli (APC) gene on chromosome 5 [1]. Approximately 25% of FAP patients carry de novo mutations and are discovered by colorectal symptoms or extracolonic manifestations [2,3]. The diagnosis is clinically based upon the presence of more than 100 adenomatous

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doi:10.1016/j.jpedsurg.2009.06.017

colorectal polyps at endoscopy [4]. Polyps begin to appear in childhood and adolescence at an average age of 16 years [2,4]. Annual sigmoidoscopy is recommended starting at the age of 10 to 12 years or from the time of diagnosis [2,4-6]. Colorectal cancer is reported at an average age of 39 years [4]. To prevent malignant degeneration, resection of the affected bowel must be performed. The timing and type of surgical intervention have to be chosen. Surgical options for FAP include total colectomy with ileorectal anastomosis (IRA), proctocolectomy with ileal-pouch anal anastomosis (IPAA), or end ileostomy. Ileorectal anastomosis is a relative simple procedure, with little complications and good functional results regarding bowel frequency and continence [2,7-9]. Furthermore, the risk of postoperative sexual and urinary problems is nearly absent. Because the remaining rectum is still at risk for the development of cancer, postoperative endoscopic surveillance is needed after surgery. The procedure should be reserved for patients with less than 5 to 20 small rectal adenomas [4,10-13]. Ileal-pouch anal anastomosis is technically a more complicated operation with a higher morbidity. In the past, the main concerns were a high frequency of bowel movements, leakage at the stapled anastomosis, and whether to use a deviating ileostomy. Despite this, the quality of life after IRA and IPAA appears to be similar [8,14]. Nowadays, IPAA is recommended in most FAP patients because it removes the colorectal cancer risk almost completely [2,7,15-17], and the follow-up surveillance of the pouch and anastomosis is relatively simple. However, a possible decreased fertility after an ileoanal procedure needs to be considered, specifically in young patients [18]. Identification of specific germ line APC or MYH mutations can aid the choice of surgical treatment [9,19,20].

The aim of this observational retrospective analysis was to evaluate the surgical outcome in young FAP patients of the Emma Children's Hospital AMC in Amsterdam.

1. Patients and methods

1.1. Patients

Between January 1977 and August 2005, a total number of 43 patients with FAP were surgically treated before the age of 26 years. The mean age was 17 years (range, 7-25 years). The male-to-female ratio was 19:24. In 34 patients, IRA was constructed and in 9 patients, IPAA.

1.2. Data collection

Before data were extracted, definitions and descriptions of variables and outcomes were defined. Data were collected using patient charts, office notes, endoscopic data (presence of <10 or >10 rectal adenomas), and surgical reports. All available resected specimens were reevaluated by the pathologist. The degree of dysplasia was described according to the Vienna classification, as low grade or high grade [21,22].

1.3. Follow-up

Early and late complications were scored respectively as being present before or after 30 days postoperatively. We scored pelvic sepsis, intestinal obstruction, anastomotic stricture, pouch fistula, pouch failure, and pouch prolapse. All complications were scored once, and in case of related complications, only the most serious one was mentioned. Data on additional surgical procedures, mortality, and the presence of ileal pouch adenomas or carcinoma of the colon or rectum were obtained.

1.4. Questionnaire

By telephone questionnaire, fecal and urinary continence, fertility, and sexual function were evaluated, using the incontinence score and diary card of Vaizey et al [23] and definitions used by Hueting et al [24], Gorgun et al [25], and Johnson et al [26]. In the Vaizey incontinence score [23], different aspects related to continence for stool and possible alterations in lifestyle are scored, related to the frequency of occurrence. The lower the score, the better the continence.

Mild fecal incontinence was defined as soiling or spotting in underwear, *severe fecal incontinence* as regular severe leakage, *fecal loss* or passive fecal incontinence, and *urge fecal incontinence* as the inability to defer defecation more than 15 minutes after the first urge.

Urinary incontinence was defined as any form of stress or urge incontinence not present before the operation.

Infertility was defined as failure to become pregnant during 12 months of unprotected intercourse without conception, with the patients being between 18 and 44 years of age and married or cohabiting. *Subfertility* was defined as successful pregnancy after having experienced periods of infertility, with or without the use of fertility treatments.

Sexual dysfunction was defined as dyspareunia, erection disorders, or retrograde ejaculation. The quality of the semen was not taken into account.

The date of last contact in the outpatient clinic, including the date at which the questionnaire was answered, was taken as the end point of follow-up.

1.5. Statistics

To evaluate statistical significance, Mann-Whitney *U* test and Fisher's Exact test (2-sided) were used to evaluate the results. A *P* value less than .05 was considered statistically significant.

2. Results

2.1. Patients

Of the 43 patients, 37 (86%) had a positive family history. The youngest patient in whom endoscopy was performed