Malignant Familial Adenomatous Polyposis treated by Laparoscopic Colectomy and Ileal Pouch Anal Anastomosis: a Case Report

Florin Zaharie1,2, George Ciorogar1,2, Roxana Zaharie2,3, Marcel Tantau2,3, Cornel Iancu1,3, Lucian Mocan1,3

1) 3rd Surgical Clinic, Iuliu Hatieganu University of Medicine and Pharmacy; 2) Prof. Dr. Octavian Fodor Regional Institute of Gastroenterology and Hepatology; 3) 3rd Medical Clinic, Iuliu Hatieganu University of Medicine and Pharmacy, Cluj-Napoca, Romania

Address for correspondence:
George Ciorogar
3rd Surgical Clinic, Iuliu Hatieganu University of Medicine and Pharmacy, Str. Croitorilor no.19-21 400162 Cluj-Napoca Romania ciorogar.george@yahoo.com

ABSTRACT

The mean age of colorectal cancer in untreated familial adenomatous polyposis (FAP) is 39 years. We present the case of a 21-year-old patient with FAP and colorectal cancer. The patient was detected with significant family history: her mother died at age 45 with colon cancer; two uncles were diagnosed with colon cancer at the age of 40 and 43 and one aunt at the age of 45 with colon cancer and gastric cancer. The treatment was laparoscopic restorative proctocolectomy with total excision of the mesorectum and ileal pouch anal anastomosis completed with endoanal excision of inferior rectal polyps. The histopathological report described a well differentiated rectal adenocarcinoma T1N1aMx developed on a tubulo-villous adenoma located on the rectosigmoid jionction, the rest of the polyps with benign histology.

Key words: familial adenomatous polyposis – laparoscopic proctocolectomy – malignant polyposis – J pouch.

INTRODUCTION

Familial adenomatous polyposis (FAP) is an inherited colorectal cancer (CRC) syndrome, characterized by the early onset of hundreds to thousands of adenomatous polyps in the colon and rectum. The lifetime risk of CRC in FAP syndrome is 100%. Colorectal cancer can be prevented by the identification of the high risk population and by the timely implementation of rigid screening programs [1, 2]. The disease occurs in about 1/10,000 to 1/30,000 individuals and accounts for less than 1 percent of all CRGs in the United States [3].

Familial adenomatous polyposis is a troublesome autosomal dominant disease that evolves quietly until clinical manifestations appear. Polyps develop beginning with puberty and gradually increase in growth until the whole colon is involved in adulthood [4].

Open restorative proctocolectomy with ileal pouch anal anastomosis (IPAA) can be performed in young patients with an acceptable functional outcome, but at the cost of relatively high complication rates, poor body image and cosmetics, and a high rate of sexual dysfunction in women. Because young patients undergoing this surgical procedure may experience negative long-term effects, surgeons should be aware of all potential consequences [5].

Long-term results after laparoscopic IPAA have been thoroughly evaluated. Laparoscopic IPAA confers excellent functional results with faster return of flatus, faster assumption of a liquid diet, and less blood loss [6].

CASE REPORT

A 21-year-old female patient was admitted in our clinic with bloody diarrhea, flatulence, anorexia, which began one month before and weight loss 10 kilos in the last 3 months. The patient had a substantial family history of CRC. Her mother died at age 45, two uncles were diagnosed with colon cancer at the age of 40 and 43, one aunt was diagnosed at the age 45 with colon cancer and gastric cancer. At clinical examination we found skin pallor and diffuse abdominal pain on palpation. The rectal examination revealed 4 polyps with 5-10 mm diameter in the anal canal.

Laboratory analysis confirmed the presence of anemia (hemoglobin 9.9 g/dL). The colonoscopy revealed hundreds of sessile polyps with a diameter of 10-20 mm in the colon, caecum, rectum and at gastroscopy numerous gastric polyps.
with a diameter of 8-9 mm were found. The case was interpreted as FAP and gastric polyps. Based on the colonoscopy aspect, malignancy was suspected. The genetic testing was not available at that moment in our clinic. We performed pelvic and abdominal CT to exclude metastasis. The pre-operative preparation of the patient consisted of enteral nutrition for five weeks, parenteral nutrition for one week and intravenous antibiotic treatment for one day. The treatment consisted of laparoscopic restorative proctocolectomy with total excision of the mesorectum and IPAA completed with endoanal excision of inferior rectal polyps.

The postoperative course was favorable, with resumption of the bowel transit and the peritoneal drains were removed on the third and fourth postoperative day. The preoperative antibiotic treatment was stopped on the fourth day, the patient being discharged on day 20 after surgery, having 6-7 continent stools / 24 hours. The morphopathological examination diagnosed a well-differentiated rectal adenocarcinoma T1N1a (Figs. 1, 2) developed in a tubulo-villous adenoma located at the rectosigmoid junction (Fig. 3). The rest of the polyps were without dysplasia. The patient followed chemotherapy: 5 cycles of 5-fluorouracil and oxaliplatin.

She presented for endoscopic assessment (gastroscopy, rectoscopy and endoscopic videocapsule) at 10 months after surgery. Examinations highlighted 10-15 hyperplastic gastric polyps with a diameter of 8-10 mm and 4-5 tubular adenomas in the rectum, with a diameter of 5-6 mm. Videocapsule endoscopy did not reveal pathological lesions. The patient was scheduled for endoscopy re-assessment after 1 year.

**DISCUSSION**

The classic form of FAP typically develops in the second or third decade of life. The mean age of polyp emergence is 16 years, but polyp onset has been noted in patients between the ages of 8 to 34 [7].

The increased risk of CRC in FAP is due to the vast number of adenomas that occur at an early age; over time, one or more of these adenomas invariably progress to CRC. Familial adenomatous polyposis is a disease of young adulthood and it can be observed before 10 years of age. Screening of clinically asymptomatic relatives thus generally starts during the teenage years. A review of 30 children with FAP and early polyp onset showed that the disease can occur in 5-year-old patients. Patients with FAP should undergo very early endoscopic surveillance. Because of the high risk of cancer in classical FAP, total colectomy is recommended if accepted after the age of puberty [8]. A review of 12 patients with FAP with the age at presentation of 7 up to 18 years showed that in one patient rectal cancer in situ was identified and a high proportion of patients had duodenal adenomatous lesions [9].

A variety of factors, including APC genotype, environmental factors, and the effect of modifier genes influence the variability of adenoma and CRC in FAP and partially explain CRC development before the age of 20. Reports from Denmark, Japan and Germany involving 2,505 FAP patients show an incidence of CRC already in early childhood, with a cumulative CRC risk exceeding 1% by 20 years of age [10, 11].

In individuals with 10 to 20 adenomas in their lifetime, the genetic testing for FAP should be considered [12]. A positive test result allows optimal surveillance and prophylactic surgery [5]. There is consensus that genetic testing in FAP is indicated when there is direct medical benefit. Therefore, when manifestations occur at pediatric age and when proven prevention strategies are in place, genetic testing for FAP in minors should be considered [13].

Patients with APC mutation are recommended to adhere to colonoscopic screening from the age of 10–12 years. Genetic testing is considered relatively uncontroversial because such
tests escape the potential for psychosocial harm, and because concerns about possible harm are overridden by the medical benefit conferred by the test [14].

The experience in laparoscopic bowel surgery has gradually increased since the introduction of laparoscopic procedures in colorectal surgery. Improved laparoscopic skills and new instruments have led to broad application in benign and malignant diseases [15]. Laparoscopic restorative proctocolectomy with IPAA is carried out in major surgical centers for elective surgery for FAP. Short and long-term results are comparable to open surgery. Proctocolectomy with IPAA is the preferred surgical option for the treatment of FAP. Low morbidity rates, good functional outcome and quality of life have been reported with open surgery [16].

Laparoscopic surgery could be an important step in FAP treatment. The high feasibility of laparoscopic colectomy for the prophylactic treatment of FAP has been demonstrated. The appeal of laparoscopic surgery could enable surgical intervention at an earlier age and lower the risk of carcinoma in patients who undergo FAP surgery [17]. The results of a study including 433 patients with a mean age of 18.04 ± 2.9 years with restorative proctocolectomy IPAA in a high volume center and increased experience with open surgery suggest that IPAA is feasible and safe in the pediatric age group and is associated with acceptable long-term outcomes [17].

Monitoring of the remaining rectum or pouch continue for the rest of the patient's life after the colon has been removed, because polyps will continue to form in the anal canal or the pouch after surgery [18]. Lifetime surveillance of the rectal stump is crucial, as shown by many authors [19]. Our patient presents gastric polyps, and their supervision by endoscopy and biopsy should be performed annually. Polyps occur in the upper gastrointestinal tract in 30 to 100% of patients with FAP [20]. Fundic gland polyps are found in most patients with FAP [21]. Dysplasia occurs in nearly half of the fundic gland polyps, although they rarely progress to carcinoma [22].

Our patient was re-assessed at 10 months after surgery, having a number of continent stools of 3/24 hours. Results using a Short Form 36 Health Survey showed that more than 7 bowel movements during the daytime and 2 defecations per day and literature review. C.I. reviewed the manuscript for its intellectual content. Z.F., C.G. and L.M. operated the patient and wrote the manuscript. R.Z. performed the endoscopies. C.I. reviewed the manuscript for its intellectual content.

Conclusions: This paper was published under the frame of European Social Fund, Human Resources Development Operational Programme 2007–2013, project no. POSDRU/159/1.5/S/138776.

REFERENCES


