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The surgical management of familial adenomatous polyposis in Northern Ireland

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SUMMARY

Sixty-eight patients from 18 families have been identified as having familial adenomatous polyposis during the past 30 years in Northern Ireland (population 1·5 million). Six of the 18 probands (33%) had developed colonic carcinoma when first seen at mean age 34 years. Ten of the 44 patients identified by surgical screening (21%) at a significantly lower mean age of 23 years had colonic carcinoma. Surgical management has generally been by subtotal colectomy with ileorectal anastomosis, or by panproctocolectomy and ileostomy.

INTRODUCTION

Familial adenomatous polyposis (previously termed familial polyposis coli), is an autosomal dominant condition characterised by the development of more than 100 adenomatous polyps in the large bowel.[1] It is associated with numerous extracolonic manifestations including osteomas, epidermoid cysts, desmoid tumours, retinal lesions, gastroduodenal polyps and adenocarcinomas, and dental anomalies. If untreated, carcinomatous changes inevitably develop in one or more of the colorectal polyps.[2] The recognition of the inherited nature of this condition has permitted an effective screening programme which can reduce the incidence of carcinoma when combined with appropriate surgery. We present a review of the surgical management of patients with familial adenomatous polyposis and of those members of their families at risk.

PATIENTS AND MATERIAL

In Northern Ireland, a total of 68 patients from 18 families have undergone surgical procedures for familial adenomatous polyposis in the past 30 years. Operative details were verified from hospital records, and, where this was not possible, by discussion with the patient or next of kin. Information was available on sixty-five of these patients.

RESULTS

Age at diagnosis was available on the 18 probands (the first member of each family to be diagnosed). The median age was 34 years (range 8 – 55 years).

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From the 18 families, 47 relatives were detected by screening, median age 23 years (range 8–43). The difference between the two groups was significant (Mann-Whitney U test: $p = 0{\cdot}008$). (Table).

TABLE

Median age, range and incidence of carcinoma in probands and patients identified by screening

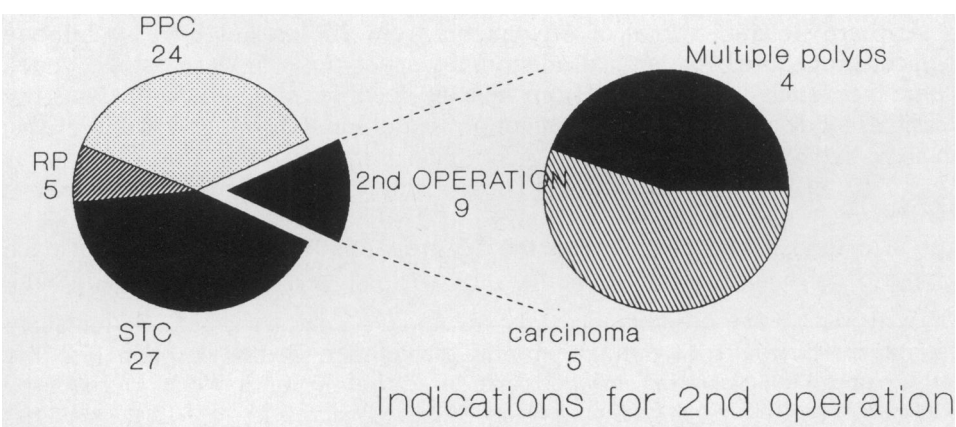
	Median age at diagnosis	*Range*	*Incidence of carcinoma*
Probands (N = 18)	34	8–55	33%
Screened (N = 47)	23	8–43	21%

A carcinoma was present in six of 18 probands when first seen and in ten of 44 patients identified by screening. This difference was not significant ($X^2 = {\cdot}75$, 1df, $p = 0{\cdot}4$). Interestingly, three patients in the screened group had been seen on one occasion before age 20 years by a medical practitioner, and because no polyps were noted, were reassured and discharged. One of these became symptomatic with multiple polyps and a carcinoma some 15 years later, and two of this patient's siblings also were shown to have carcinomas. Only one child of an affected individual had a carcinoma, out of 29 found to have polyps on screening. When compared with the probands, the difference in incidence of carcinoma was significant ($X^2 = 7{\cdot}826$, 1df, $p = 0{\cdot}05$).

Thirty-six patients were treated by subtotal colectomy and ileorectal anastomosis, nine of whom subsequently required abdominoperineal excision of the rectum. (Figure). The indication for excision of the rectum in five patients was carcinomatous changes in the rectal segment, and in four extensive 'carpeting' with

FIGURE

Surgical procedures for familial adenomatous polyposis



PPC = Panproctocolectomy. RP = Restorative proctocolectomy.
STC = Subtotal colectomy/ileorectal anastomosis.

polyps. The mean period of follow-up to malignant transformation was 12 years. In this group two patients died from metastatic disease within two years of diagnosis of their rectal tumour. The remaining three patients are alive and well two to ten years after rectal excision. The median age for probands treated by colectomy and ileorectal anastomosis was 35 years (range 11–54 years). For those detected by screening and treated in this manner the median age was 19 years (range 8–37 years).

Twenty-four patients were treated by panproctocolectomy and ileostomy formation, of whom fifteen had a carcinoma at time of presentation. Nine patients had multiple polyps but no evidence of malignancy. The mean age at time of diagnosis for the probands was 39 years (range 24–55 years) and for those screened 31 years (range 16–42 years). Five patients have been treated by restorative proctocolectomy, median age at diagnosis 25 years (range 17–40 years). Five patients have undergone a variety of surgical procedures including colectomy, laparotomy alone (when carcinomatosis was found) and fulguration.

DISCUSSION

The first description of familial adenomatous polyposis is attributed to Harrison Cripps in 1882.[3] For many years the surgical treatment was restricted to local excision of large polyps which could be removed *per rectum,* or to palliative procedures for patients who had developed carcinoma. Colectomy for the control of established disease was not considered until after 1925, but was associated with an extremely high post-operative mortality, figures of 25% being quoted for abdominoperineal excision of the rectum and 50% for colectomy.[4] More recent overall figures for the morbidity and mortality associated with large bowel resection for malignant disease have been estimated to be approximately 9% and 13% respectively.[5] It is clear that the morbidity and mortality associated with ileorectal anastomosis performed as an elective procedure in a young fit population would be significantly lower. In our own practice, operative mortality for elective colonic surgery in this age group is virtually zero, and wound and intraperitoneal sepsis have occurred in less than 2% of cases.

In 1939 McKinney described panproctocolectomy with ileostomy formation, or, as an alternative, subtotal colectomy and ileosigmoid anastomosis, with follow-up fulguration of polyps.[6] In 1957 Hubbard noted regression of rectal polyps after subtotal colectomy and ileorectal anastomosis, an observation which helped to popularise this operation.[7] The ideal procedure should remove all large bowel mucosa yet preserve normal or near normal bowel function, and thus avoid a stoma. The operation should have an acceptable level of morbidity and preserve normal sexual function.[6]

At present there are three options available to the surgeon: i) panproctocolectomy with ileostomy, ii) subtotal colectomy and ileorectal anastomosis, and iii) restorative proctocolectomy. No single procedure is suitable for every patient. The desires of the patient, the preference of the surgeon and the proposed follow-up must all be considered before a decision is reached. Panproctocolectomy with Brooke ileostomy has the advantage of being a straightforward procedure, with a relatively low perioperative complication rate. In those presenting with a carcinoma in the middle or lower rectum it is the operation of

choice, and may be considered the best option in the elderly or debilitated patient.[10] The disadvantages of this operation include cosmetic and psychological problems associated with the stoma, and the risk of sexual or bladder dysfunction if a close rectal dissection has not been undertaken. In addition, there are a number of complications associated with ileostomy formation including skin irritation, retraction, prolapse, and parastomal hernia. Panproctocolectomy removes the entire colonic mucosa with its malignant potential. In the past this was considered a curative procedure with routine follow-up being unnecessary, but the recognition of upper gastrointestinal polyps, and malignancy in duodenal polyps, has highlighted the need for long-term review.[9,11] The reported incidence of upper gastrointestinal polyps ranges from 24–100% in patients affected by familial adenomatous polyposis, and there are no clear guidelines on management.[9,12]

The risk of malignancy developing in the duodenum of patients with familial adenomatous polyposis is 50–100 fold greater than that of the general population; however, the risk is still small. At present surgeons are reluctant to recommend a Whipple's procedure without evidence of frank malignancy, but it is advisable to perform regular upper gastrointestinal endoscopy and biopsy of polyps. Large polyps may be removed by snaring or fulgurated as in the rectum, but clearance of all duodenal polyps is seldom feasible.

Ileorectal anastomosis has been the most popular operation in this condition. The operation is safe and the sphincters and the pelvic autonomic nerve plexuses remain intact. Following colectomy and ileorectal anastomosis it is necessary to deal with the largest rectal polyps by fulguration or snaring, and the patient must attend for regular follow-up with proctosigmoidoscopy. Polyps of 5 mm or more should be removed or fulgurated, but it is unnecessary and indeed may be inadvisable to remove all minute polyps. Repeated fulguration of the rectum can lead to extensive scarring of the rectal mucosa, so that early malignant transformation may be difficult to recognise. Follow-up examination is performed every six to 12 months, and polypectomy is required on average every two to three years.[13] The long-term risk of malignant transformation in the rectal remnant varies greatly, from as low as 13% in the St Mark's series to as high as 59% in the Mayo Clinic series.[14,15] Where numerous polyps exist or where they form almost a complete 'carpet' in the rectum, rectal excision should be considered. Even if the patient attends regularly for follow-up there is no guarantee of protection from carcinoma formation. Indeed we report two cases where tumours developed in spite of regular follow-up.

The risks of rectal cancer must be considered by both surgeon and patient when deciding on the operation to be performed. The ileorectal anastomosis ensures good bowel function, has a low complication rate and simple follow-up, but patient compliance is vitally important. The importance of this follow-up must be impressed on the patient, and if there is a possibility that the patient would be unco-operative another procedure should be considered. We advise combined upper and lower gastrointestinal flexible endoscopy at a single visit on an annual basis in established disease involving upper and lower gastrointestinal tracts, with a rigid sigmoidoscopy once in the interval. If no upper gastrointestinal manifestations are present then oesophagogastroduodenoscopy every three years is probably adequate.

Restorative proctocolectomy with pouch formation and ileoanal anastomosis appears to be closest to the ideal surgical procedure for patients with familial adenomatous polyposis. In this operation, all colonic mucosa is removed, near-normal bowel function can be preserved with avoidance of stoma formation, sexual function is preserved and in specialist units the complication rate is low.[16] Complications nevertheless are more frequent than with other pelvic resections, especially pelvic sepsis. In 5–6% of cases the procedure fails, and a permanent ileostomy is needed. A defunctioning ileostomy is required until the ileoanal anastomosis has healed. "Pouchitis" has now been recorded in cases of restorative proctocolectomy.[17] Unfortunately frequency of defaecation and anal leakage may mar the results of surgery and this operation cannot therefore be considered the procedure of choice in most cases.

The advantages of screening the offspring of these patients are reflected in the reduced incidence of carcinoma at time of diagnosis. To identify those at risk it is necessary to have accurate pedigrees. Those at risk can be advised to attend for screening. The use of a polyposis register has been shown to reduce the incidence of carcinoma in those carrying the gene for familial adenomatous polyposis.[18, 19] As part of ongoing research into gene markers of this condition we have been collecting information on families.

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