Polyposis Coli
The Clinical Spectrum in Adults

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SUMMARY
Four patients are reported, to illustrate the wide spectrum of colonic polyps which may occur in multiple form in adults. Included are unusual cases of metaplastic and juvenile polyposis. The subject is reviewed, and the need for accurate histological diagnosis is stressed. The possibility that the difference between familial adenomatous polyposis and Gardner's syndrome may lie merely in the degree of expressivity of the associated extracolonic features is discussed. Surveillance of these patients for peri-ampullary neoplasms in the duodenum is considered. The interrelationship between familial adenomatous polyposis, juvenile polyposis and colorectal cancer is discussed.


A variety of polyps occur in the colon and rectum in multiple form in the adult. Although none is common, some are well documented and of particular interest because they are inherited. These are: familial adenomatous polyposis (adenomas), Gardner's syndrome (adenomas), juvenile polyposis (hamartomas), and Peutz-Jeghers polyposis (hamartomas). A second miscellaneous group includes the common metaplastic polyps, those arising in lymphoid tissue (benign and malignant lymphoid polyposis), and the inflammatory pseudopolyps or mucosal tags associated with inflammatory bowel disease. Rarely, polyposis coli may represent an unusual manifestation of a variety of conditions such as neurofibromatosis or lipomatosis, and occasional examples of the Cronkhite-Canada syndrome and Turcot syndrome have been recorded.

Polyposis coli has a low incidence. The commonest form, besides multiple small metaplastic and lymphoid polyps and adenomatous polyps, is familial adenomatous polyposis, the incidence of which has been estimated at only 1/24 000 births in Britain. Patients with multiple polyps are thus sufficiently unusual for knowledge regarding the clinical varieties and their natural history to emerge only slowly.

In this article 4 patients are described to illustrate unusual features of different types of polyposis, and to stress the need for exact histological diagnosis. In addition, two areas of special interest are discussed: the relationship between familial adenomatous polyposis and Gardner's syndrome, and the interrelationships between the inherited polyposis syndromes and colorectal carcinoma.

CASE REPORTS
Patient 1
A White man aged 42 years presented with a 4-month history of mucous diarrhoea. Sigmoidoscopy disclosed 20 stalked polyps up to 25 cm from the anal verge. The histological appearance of one, removed by biopsy, was that of a metaplastic polyp. The majority of these polyps were 0.75 -1.5 cm in size. Air contrast barium enema examination (Fig. 1) and colonoscopy showed further polyps up to the splenic flexure, the total number being approximately 30. Twenty of these were removed by colonoscopic polypectomy, with resultant symptomatic improvement. Histological examination confirmed the nature

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of the metaplastic polyps (Fig. 2). Barium studies failed to reveal upper gastro-intestinal pathology, and there was no family history of colonic disease.

Fig. 2. Patient 1. The typical histological features of a metaplastic polyp. All polyps had the same structure.

Patient 2

A White man aged 41 years presented with a 2-week history of rectal bleeding. His brother had undergone surgery for a colonic carcinoma at the age of 45 years. Sigmoidoscopy and colonoscopy disclosed 15 stalked polyps to the mid-descending colon, all of which were removed by colonoscopic polypectomy. The remaining mucosa was normal. The polyps were between 0.5 cm and 1 cm in size and showed the typical histological features of juvenile polyps (Fig. 3).

Fig. 3. Patient 2. The histological appearance of one of the multiple juvenile polyps.

Patient 3

A White woman presented at the age of 57 years with signs of peritonitis due to a perforated carcinoma of the ascending colon. A right hemicolectomy was performed, the specimen showing multiple adenomatous polyps in addition to the carcinoma (Dukes grade C). After recovery, colonoscopy demonstrated numerous polyps in the remaining colon and rectum, 3 of which showed adenocarcinoma histologically. Total colectomy with ileorectal anastomosis was performed and the rectal stump cleared of polyps with diathermy. There were more than 250 polyps in the removed specimens. No member of the patient’s family had suffered from colonic disease, and 3 siblings aged between 18 and 28 years have so far declined investigation.

Patient 4

A young girl from a family known to be prone to polyposis, whose mother died from carcinoma of the colon, had had adenomatous polyposis coli at the age of 10 years. When aged 18 she agreed to total colectomy and ileorectal anastomosis. Investigation before surgery showed multiple osteomas in the mandible, but no other lesions.

DISCUSSION

Metaplastic (Hyperplastic) Polyposis Coli

Smooth mucosal nodules are frequently seen as an incidental finding on sigmoidoscopy. Biopsy will confirm the benign nature of the metaplastic polyp. Usually 1 - 2 mm in size, these small nodules are found in the rectum in about 80% of patients over 40 years of age, and are no more common in patients with large bowel carcinoma, and have no malignant potential. However, occasionally they can be large, stalked and multiple, and can thus resemble adenomatous polyps, as in patient 1, in whom the number and size of the polyps were exceptional. Metaplastic polyps only rarely give rise to symptoms, and removal by means of the colonoscopic snare proved effective and safe in this case.

Juvenile Polyposis Coli

Juvenile (retention) polyps usually occur singly or as a few lesions. The average age of patients who present with these lesions is 6 years, and 80% occur in children below 10 years of age. Histologically they are hamartomas, consisting of dilated epithelial tubules embedded in an excess of lamina propria. Occasionally they occur in multiple form in the colon, and less commonly may be found in other parts of the gastro-intestinal tract. They usually present with bleeding or prol. pse per rectum, and auto-amputation is common.

Rarely the multiple form may occur in adults, as in patient 2. The similarity to the adenomatous form of polyposis explains why such patients were frequently treated by colectomy before the true nature of the juvenile polyp was appreciated. The possible relationship between juvenile polyposis, familial adenomatous polyposis and colorectal carcinoma is discussed below. The occurrence of colonic carcinoma in this patient’s brother at a younger age may have a genetic basis.
Familial Adenomatous Polyposis Coli

In this precancerous, genetically transmitted condition, multiple adenomatous polyps are present in the colon, varying from about 100 to many thousands. It has been suggested that when more than 100 adenomatous polyps are present the condition is one of major or familial polyposis coli, inherited through a dominant gene, 50% of the offspring being affected. In the so-called recessive or minor adenomatous polyposis, on the other hand, up to about 100 adenomatous polyps may be present in the colon, usually with no family history. Whether a recessive gene is responsible for the latter is as yet not clear. If patient 3 is presumed to have major adenomatous polyposis, this must have arisen by spontaneous mutation in the absence of a family history. This occurs in one-third of affected patients with this disease.

The mean age of presentation is 22 years, and most patients develop cancer by the age of 40 years. However, patients who presented at 4 months and at 74 years of age have been reported. Although cancer may rarely develop in patients younger than 16 years of age (11 reported cases to 1972), preventive surgery is usually advised as soon after puberty as the diagnosis is made. At the other end of the scale, the late age of presentation (over 50 years in the case of patient 3) is characteristic of 1 in 8 patients with this disease seen at St Mark’s Hospital in London, indicating that surveillance throughout life is necessary. Sixty-seven per cent of those patients who present with symptoms already have cancer, compared with an incidence of 10% in those relations who are investigated because they are at risk for polyposis coli.

The most widely accepted prophylactic treatment is colectomy with ileorectal anastomosis, accepting that the rectal stump remains at risk for the development of cancer. This risk is small — 3.6% at 20 years follow-up according to the St Mark’s Hospital figures. In sharp contrast to this, the results from the Mayo Clinic suggest an incidence of 42% at 20 years. However, subsequent assessments by other centres in the USA have failed to demonstrate a similar high incidence. An additional drawback of ileostomy is that the fear of a stoma may lead relations who are at risk to decline investigations or treatment.

After ileorectal anastomosis, regression of the remaining rectal polyps has been observed. However, further polyps have been noted to develop in such patients who have been followed up for a long period.

Gardner’s Syndrome

Gardner originally described patients with inherited adenomatous polyposis in whom epidermoid cysts, soft-tissue tumours and osteomas of mandible or skull were present, not necessarily all in the same patient. The colonic manifestations of this syndrome appear to be very similar to those of familial adenomatous polyposis in terms of number, distribution, age of onset and natural history of the polyps, and the penetrance of the gene is high in both conditions with regard to the polyposis (about 90%). However, the expressivity of the extracolonic manifestations of Gardner’s syndrome is variable, and its frequency is probably underestimated among patients with polyposis. In fact, extracolonic manifestations were found in 93% of 29 patients with familial polyposis coli and no obvious stigmata of Gardner’s syndrome who were specifically investigated for osteomas in the mandible.

The same finding occurred in patient 4. This raises the question as to whether the difference between the two conditions is merely one of variation in the expressivity of the different extracolonic manifestations associated with the polyposis. Should this be the case, it becomes of practical consequence in view of a number of other extracolonic features which have been described subsequent to Gardner’s original papers. These include dental anomalies, dermoids, and keloid scars. In particular, peri-ampullary malignancy and polyps in the duodenum are being reported with increasing frequency in patients with inherited adenomatous polyposis with or without other extracolonic stigmata. Many families with polyposis have been reported in which some members have been shown to have extracolonic abnormalities, while in other members these have not been demonstrated. It would seem that there is sufficient evidence to suggest that all patients with inherited adenomatous polyposis coli may be at risk to develop extracolonic abnormalities, and that the upper gastrointestinal tract in particular should be investigated in these patients.

Interrelationships between the Polyposes

Occasionally the adenomatous polyps of familial polyposis coli may be preceded by, or mixed with, metaplastic polyps and, rarely, juvenile polyps. Juvenile polyposis has also been reported more and more frequently to occur in families. Of more relevance, juvenile polyposis, familial adenomatous polyposis and carcinoma of the colon may sometimes occur in the same family. Veale et al. reported 11 patients with juvenile polyposis who came from 8 families. Four of these were particularly interesting. In 2 of these families, the offspring of fathers who had carcinoma of the colon developed juvenile polyposis coli. In 1 of these families 2 subsequent sibs inherited the disease. In the third family 1 of 2 brothers with familial polyposis coli had a daughter who developed juvenile polyposis. In the final family, 4 generations were involved. Father and son had colon cancer, and successive direct descendants suffered from familial adenomatous polyposis and juvenile polyposis. Despite this and other reports, however, the rarity of these conditions has to date prevented complete clarification of the genetic relationships between them. It would seem that the tendency to develop carcinoma of the large bowel is clear in individuals who inherit familial polyposis coli, and remains a possibility in the descendants of those with juvenile polyposis coli.

CONCLUSIONS

This report illustrates the wide spectrum of colonic polyps which may occur in multiple form in adults.
Correct management depends upon accurate histological diagnosis, which ranges from the benign to the inevitably malignant. The relationship between familial adenomatous polyposis and Gardner's syndrome is discussed and it is concluded that it is reasonable to consider all patients with inherited adenomatous polyposis to be at risk for developing extracolonic manifestations, particularly malignancy in the peri-ampullary region. The link between familial adenomatous polyposis, juvenile polyposis coli and colorectal cancer is discussed, and besides the known risk of malignant change in adenomatous polyposis, there is evidence to suggest that surveillance should be maintained on the descendants of those with juvenile polyposis, since they may be a high-risk group with regard to the development of carcinoma of the colon.

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REFERENCES


Value of the Mandibular Radiograph in Familial Polyposis Coli

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SUMMARY

Familial polyposis coli and Gardner's syndrome have identical colonic manifestations. Recent evidence has shown that both conditions are associated with occult osteomatous lesions in the mandible. We present 3 patients from 2 families with familial polyposis coli who, in addition to the colonic lesions, also had occult osteomatous mandibular lesions. The value and practical application of radiography of the mandible as an additional diagnostic aid in familial polyposis coli are emphasized.

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Familial polyposis coli (FPC) is a rare disorder with an estimated incidence of 1 in 8 000 in the general population. It is inherited as a Mendelian autosomal dominant trait. Although some cases have appeared to be sporadic, most genetic studies have found the penetrance of the gene to be approximately 95%. It is possible that the occasional patient without a family history reflects failure to detect the disease in his relatives. Thus, the diagnosis of FPC carries the need to search for the lesions in other family members, and to offer genetic counselling to affected prospective parents.

Carcinoma of the colon is inevitable in untreated cases of FPC. The carcinoma usually develops some 15 years after the appearance of the colonic polyps. In one large series, the average age at which polyps appeared in the colon was 25 years.

Sigmoidoscopy and barium enema examination are used in the detection of polyps in patients suspected of having FPC. Recently, the value of radiography of the mandible...