



POLYP

A polyp is a localised elevated lesion arising from an epithelial surface. If it has a stalk it is called a pedunculated polyp whereas a sessile polyp is broad based and lacks a stalk.

Polyps are not necessarily neoplastic but may be hamartomas, or secondary to inflammation. Table 1 lists the more common polyps and the conditions associated with them.

ADENOMA

An adenoma is a benign epithelial neoplasm, the majority (75%) of which are tubular. In these, the abnormal epithelium is the dark red head, which may be round, oval or lobulated. The stalk consists of normal epithelium. A villous adenoma is usually sessile and accounts for 10% of adenomas. It has a broad base and a shaggy surface. Tubular villous adenomas have both tubular and villous features and account for 15% of all adenomas. The macroscopic forms of the adenomas bear little relationship to the microscopic appearance. The histological appearance varies from branching tubular glands to finger like elongated villi. All adenomas may show different histological patterns in different areas. The degree of differentiation decreases as the polyp becomes more like cancer. The presence of severe atypia is highly suggestive of malignancy in the epithelium of the polyp, where direct evidence of invasion of the muscularis mucosa has not been identified. Once invasion of the muscularis mucosa has occurred, the lesion is an adenocarcinoma.

POLYP-CANCER SEQUENCE

A benign polyp is the usual precursor lesion in most patients with colorectal cancer. Occasionally cancers may arise de-novo from normal epithelium, or from

lesions called flat adenomas. Evidence for the polyp cancer sequence includes:

1. Polypectomy decreases the incidence of cancer.
2. Colonic adenomas occur more frequently in patients with cancer. 30% of patients with a colorectal cancer will have a polyp elsewhere.
3. Large adenomas are more likely to have areas of cancer within them than small ones.
4. Severe dysplasia has been shown to progress to cancer in polyps.
5. Residual adenomatous tissue is found in most invasive colorectal cancers.
6. Patients with familial adenomatous polyposis develop cancer in 100% of cases. Their adenomas are the same as sporadic adenomas.
7. There is a high incidence of adenomas in populations with high rate of cancer.
8. The presence of an adenoma increases the life-time risk of colorectal cancer.

	Solitary	Multiple
Neoplastic	Adenoma: <ul style="list-style-type: none"> • Tubular • Tubulovillous • Villous • Adenocarcinoma • Flat adenoma 	Familial adenomatous polyposis
Hamartomas	Juvenile	Juvenile polyposis Peutz-Jeghers
Inflammatory (Pseudopolyp)	Ulcerative colitis Crohn's disease Dysentery's Diverticulitis Amoebiasis Bilharzia	Inflammatory polyposis in colitis
Unclassified	Metaplastic (hyperplastic)	Multiple metaplastic polyposis

TABLE 1: POLYPS AND THEIR ASSOCIATED SYNDROMES.

MALIGNANT POTENTIAL OF ADENOMAS

The vast majority of adenomas do not develop into carcinomas. However, the factors important in malignant transformation are shown in table 2. Flat adenomas have a disproportionately high rate of severe dysplasia. The life history of the adenoma - carcinoma sequence is usually greater than 5 years and averages 10-15 years. In hereditary non-polyposis colorectal cancer however, it is much shorter, in the region of 3 years.

	Malignancy rate (%)
Size:	
<1cm diameter	1
1-2cm	10
>2 cm	50
Histology:	
Tubular adenoma	5
Tubulovillous adenoma	20
Villous adenoma	40
Epithelial dysplasia:	
Mild	5
Moderate	20
Severe	40

TABLE 2: POLYP CHARACTERISTICS ASSOCIATED WITH THE RISK OF MALIGNANCY.

CLINICAL PRESENTATION

The most common presentation is with bleeding, which may be bright or dark red and may be associated with the passage of mucus. Diarrhoea is uncommon but may occur with large villous adenomas. Large polyps may prolapse through the anus.

Diagnosis

Complete examination of the large bowel is essential.

1. *Sigmoidoscopy and double contrast barium enema.* Double contrast barium enema may miss lesions because of poor preparation, or when lesions are in sigmoid colon where overlapping of bowel or diverticular disease may impair mucosal definition. Visualisation of the rectum is inadequate with a barium enema.
2. *Colonoscopy* is a more reliable way to diagnose polyps.

Treatment

Total excision biopsy of the polyp is the treatment of choice as this allows the pathologist to examine the entire lesion. The method of removal depends on the site and whether the polyp is pedunculated or sessile.

Pedunculated polyps

These are removed by snare excision through a colonoscope. Occasionally large lesions may require a laparotomy and colotomy for removal.

Sessile polyps

Small sessile lesions may be removed by snare excision through a colonoscopy, but this requires advanced techniques. Perforation of the colon is a significant risk. These lesions are therefore usually managed by colotomy.

Follow up

About half of all patients who have adenomas removed will develop new ones within 10 years. This risk is highest for patients who have multiple tubular adenomas or a villous adenoma at their initial examination. Therefore patients with the highest risk of new adenomas should probably have a repeat colonoscopy at 2-3 year intervals, while those with a low risk at 4-5 year intervals until the risk of dying from colorectal cancer is lower than the risk of dying from other diseases.

Patients who have adenomas have a 2-3 times greater risk of developing colorectal cancer than the normal population. Once all polyps have been removed however, the risk of developing colorectal cancer is reduced to below that of the normal population.

MALIGNANT COLORECTAL POLYPS

The majority of malignant polyps can be successfully treated by polypectomy alone (by snare or local excision) provided the specimen is carefully examined and that the following histopathological criteria are strictly applied.

1. The lesion is completely excised.
 2. It is well or moderately differentiated.
- No further action other than careful follow up by colonoscopy is necessary. If the resection line is involved by tumour, or the tumour is poorly differentiated, formal surgical excision of that section of the colon is indicated.

Juvenile polyp (Retention polyp)

These occur most commonly in children, less frequently in adolescents and only occasionally in adults. They are usually solitary but multiple polyps are not uncommon. They may occur in any part of the colon but are most common in the rectum. They are smooth with a rounded contour. The outer surface is often ulcerated. Numerous mucin containing cysts are visible on the cut surface. The polyps are usually pedunculated. The pedicle does not have a muscularis mucosae so auto amputation is common. Patients usually present with bleeding from the ulceration of the tip of the polyp, or as a result of auto amputation.

These polyps are thought to be hamartomatous, without malignant potential.

Juvenile polyposis

This is a rare autosomal dominant condition. Multiple polyps occur in the stomach, small bowel and colon. Patients develop anaemia or frank rectal bleeding from the colonic polyps. The small bowel polyps may cause intussusception. Cancer may develop in the colonic polyps, usually by fourth decade.

Treatment

1. Colonoscopic removal of colonic and rectal polyps.
2. Trans-abdominal polypectomy of small bowel polyps greater than 2 cm in size.

Peutz-Jeghers polyposis

The polyps in Peutz-Jeghers polyposis and hamartomas. They appear tree-like and include the muscularis mucosae in the polyp. The polyps occur throughout the gastrointestinal tract, but are most numerous in the small bowel. Colonic polyps are unusual. These patients may have the classical mucocutaneous melanin pigmentation usually seen on the lips and buccal mucosa. The pigmentation may also occur on the fingers and toes. The condition is an autosomal dominant.

Symptoms

The multiple polyps may result in minor bleeding over a long period of time resulting in chronic anaemia or a major bleed. The small bowel polyps may cause intussusception. There is an increased risk of carcinoma of the stomach and small and large bowel. There is an increased risk of non-gastrointestinal malignancies such as ovary, fallopian tube, thyroid and lung. Almost all malignancies are fatal. The average life expectancy is 40 years.

Familial polyposis coli (FAP)

This condition was one of the first inherited conditions identified that predisposed to cancer. It is uncommon, accounting for only 1% of all large bowel cancers, but is important because family members need lifelong surveillance. The disease does not only affect the colon, but often results in abnormalities in other parts of the gastrointestinal tract such as the stomach (hamartomas and adenomas), duodenum (adenomas) and to a lesser extent, the jejunum and ileum. Extra-intestinal manifestations may also occur in a variant of the condition called Gardeners Syndrome.

The condition is caused by mutation of the APC gene on chromosome 6. The mutations are family specific and the way the disease manifests depends to some extent on where in the gene the mutation occurs. An individual may either inherit the mutation in the APC gene from either parent or may develop a new mutation. Over 99% of those individuals who have the mutation will develop the colorectal cancer.

Symptoms

Patients with polyps are initially asymptomatic. They develop increased stool frequency, and then begin to pass of mucous and blood. Symptoms usually begin around 25 years of age. Once symptoms develop, a quarter will already have a colonic cancer.

Diagnosis

Rigid or flexible sigmoidoscopy is the essential examination. Any polyps should be biopsied to confirm the presence of adenomas. The rectum is virtually always affected to a greater or lesser degree. Colonoscopy is useful to detect cancers. The number of polyps varies but by definition, if there are more than 100 colorectal adenomas, the patient has FAP. In young patients with a family history of FAP, far less than 100 is adequate to make a diagnosis.

Natural history

Familial adenomatous polyposis is an autosomal dominant condition. The penetrance is in excess of 90%. Males and females are affected equally and either may transmit the disease to their offspring. Only those with polyposis can transmit it to next generations.

The colon and rectum are normal at birth. Polyps usually develop during childhood, usually about puberty. Symptoms typically develop in the mid to late twenties. Colonic cancer usually occurs in the mid-thirties and affected family members die in the early forties.

Aetiology

Mutation of the APC (adenomatous polyposis coli) gene on chromosome 5q is responsible for the disease. This gene has now been sequenced. Different families have mutations in different parts of the gene. Abnormalities in different areas of the gene are associated with different clinical manifestations of the disease.

Gardeners syndrome

This condition is familial adenomatous polyposis associated with osteomata of skull and mandible and epidermoid cyst and soft tissue tumours of skin. Other associated abnormalities include dental abnormalities such as supernumerary teeth, impacted teeth, dentigerous cyst and irregular island of increased bony density in the mandible, maxilla and skull.

Desmoid tumors

These occur mainly in operation scars in the abdominal wall, and in the mesentery of small bowel. About 10% of patients with FAP may develop them. The most dangerous variety is the intra-abdominal desmoid which may cause intestinal obstruction resulting in small bowel perforation, or result in an obstructive uropathy. Intra-abdominal desmoids are the second most common cause of death if colon cancer is prevented, being responsible for 10% of all deaths from familial adenomatous polyposis.

CONGENITAL HYPERTROPHY OF RETINAL PIGMENT EPITHELIUM (CHRPE)

CHRPE is a discrete, darkly pigmented, round, oval or kidney shaped lesion seen in the retina in affected members of some families with familial adenomatous polyposis. They were of great interest previously because they are detectable on fundoscopy at an early age before the colonic polyps developed. Their usefulness has been superseded by accurate genetic testing.

EXTRA COLONIC MALIGNANT LESIONS

Patients with familial adenomatous polyposis are at risk for malignancies in organs other than the colon. Periampullary carcinoma occurs in the late fifties or early sixties. It is the commonest cause of death once the risk of colorectal cancer is removed. Duodenoscopy every 5 years is recommended for all patients with familial adenomatous polyposis who have had a colectomy. Other tumour include:

- C.N.S. tumours (Turcot syndrome)
- Thyroid carcinoma
- Adrenal carcinoma
- Hepatoblastoma
- Gastric carcinoma

Management

a. Prophylactic surgery

The risk of developing colorectal cancer in a patient with familial adenomatosis

polyposis is in excess of 80%. Prophylactic surgery is intended to prevent the development of colorectal cancer. There are 3 operations which reduce the risk of cancer:

Proctocolectomy: The removal of the entire colon and rectum. The small bowel terminates in a permanent end ileostomy. This operation has the advantage of removing all the colonic mucosa, but results in a permanent ileostomy. Few young asymptomatic patients would find this an acceptable procedure.

Colectomy with ileo-rectal anastomosis. The whole colon is removed and the terminal ileum is joined to the upper rectum. The advantages of this procedure are that it is a relatively simple operation and can be performed by the average general surgeon. The morbidity is low and the functional results are acceptable with the passage of about 3 stools per day. The disadvantage is that rectal mucosa is left behind, and the patient is at risk for the development of rectal cancer. Six monthly sigmoidoscopy surveillance of the rectal remnant is essential. The rectum usually needs to be removed 15 to 20 years later.

Restorative proctocolectomy involves removal of the entire colon and rectum, with the construction of a pouch at the end of the small bowel. This pouch is anastomosed to the anus. The operation is more extensive than colectomy and ileorectal anastomosis with a greater risk of complications. It is only performed by specialist trained colo-rectal surgeons. The stool frequency is slightly higher than after an ileorectal anastomosis. There is a much greater risk of impotence in the male and impaired fertility in the female.

It is difficult to choose between the 2 operations. The general trend in the USA is to do a restorative proctocolectomy as the procedure of choice, while in the UK, an ileo-rectal anastomosis is usually done. A good compromise is to do an ileo-rectal anastomosis in the late teens or early twenties provided there are not many rectal polyps. If rectal polyps become a

problem, or the diagnosis is made late in the twenties, then a restorative proctocolectomy should probably be done.

b. Patients with colorectal cancer

Some patients present with colorectal cancer and familial adenomatous polyposis is identified during the pre-operative work-up. If the treatment is likely to be palliative, the cancer alone should be managed. If however, the treatment is likely to be curative, the operation of choice is a restorative proctocolectomy.

MANAGEMENT OF THE POLYPOSIS FAMILY

A family tree must be constructed. All members at risk for the disease should be examined by sigmoidoscopy starting at 13 or 14 years and continued every second year until the age of 40 years. In the occasional family where polyps and cancer occur late, sigmoidoscopy should continue until 60 years of age or even longer.

Mutation identification is now possible in many families who have familial adenomatous polyposis. This allows accurate diagnosis before the appearance of colonic polyps. There are, however, many ethical issues which make consent for genetic testing difficult, especially in children. At risk family members who do not have the mutation can be reassured

that their risk of developing cancer is the same as the general population, and they will not pass the disease on to their offspring. Those who have the mutation are at extremely high risk for the development of cancer. They need close follow-up and prophylactic colectomy once they develop colonic polyps. Individuals who know that they have the mutation have other problems that are not immediately obvious. For example, the mutation must be declared when applying for Life Insurance, which would usually be refused. Without life insurance, the individual cannot raise a mortgage, and therefore cannot buy a house. Because of these issues, it is essential that individuals at risk for diseases like familial adenomatous polyposis are carefully counselled by a qualified genetics councillor both before the test and at the time of release of the results. The individual should be of sufficient maturity to understand the implications of the test, and therefore should be at least 18 years old. It is questionable whether parents can make this type of decision for their children, especially as the test result will make little difference to management of the individual until early adulthood.



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