Bowel (colonic) polyps

Familial polyposis of the colon causes widespread development of adenomas in the colon and rectum. The number of polyps can range from no detectable polyps at colonoscopy to more than 7,000 seen on resected specimens of bowel. The polyposis predominantly affects the distal colon. [1]

Attenuated familial adenomatous polyposis (AFAP) [2]

Attenuated familial adenomatous polyposis is characterised by fewer colonic polyps (100 or less) and a delayed onset of symptoms and complications. The colonic polyps tend to involve the proximal colon and spare the rectum.

- The true incidence of attenuated familial adenomatous polyposis is not known.
- There is a delay in onset of adenomatosis and bowel symptoms of 20-25 years, a delay in onset of colorectal cancer of 10-20 years and a delay in death from colorectal cancer of 15-20 years.
- Because of the tendency to affect the proximal colon, colonoscopy is preferred to sigmoidoscopy for surveillance, which should begin at the age of 20-25 years.
- There are often associated gastric and duodenal adenomas and so regular upper gastrointestinal endoscopy is also recommended.

Gardner's syndrome is characterised by colonic polyposis with osteomas and soft tissue tumours. [3] Turcot's syndrome is the association of colonic polyposis and tumours of the central nervous system. [4] In 1951 Gardner described the occurrence of familial adenomatous polyposis with the extra-colonic manifestations of desmoids, osteomas and epidermoid cysts.

How common is familial polyposis?

- The incidence of familial polyposis of the colon is about 1 in 10,000 live births.[1]
- Colonic polyps begin developing at a mean age of 15 years.[1]
- Familial polyposis syndromes have autosomal dominant inheritance with almost complete penetrance but marked variation in expression. Mutations of the APC gene on chromosome 5 are thought to be responsible.[5]
- The location of the mutation on the gene is thought to influence the nature of the extra-colonic manifestations.

Presentation

- Adenomas usually begin to develop during the second decade of life.
- Unfortunately, it often presents with colorectal cancer.
- The median age at diagnosis is 40 years.[5]

Symptoms

- Patients are often asymptomatic but may present with rectal bleeding, diarrhoea, abdominal pain and mucous discharge.[6]
- Obstruction may cause constipation, vomiting and peritonitis.

Signs

- Rectal polyps or masses.

- Other commonly associated features:[5]
    - Congenital hypertrophy of the retinal pigment epithelium (CHRPE) - evaluated by slit-lamp examination and indirect ophthalmoscopy - which can be a useful early clue as to whether the patient is a carrier of the APC gene.
    - Dental problems - supernumerary teeth, odontomas, non-erupted teeth.
    - Epidermoid cysts.
    - Desmoid tumours or osteomas (skull, endosteal and exosteal osteomas).
    - Thyroid masses.

Investigations

- FBC.
- Carcinoembryonic antigen testing: raised levels may indicate colorectal carcinoma.
- LFTs to evaluate possible metastasis.
- TFTs.
- Faecal occult blood.
- CT or MRI scan of the abdomen and pelvis.
- Dental X-rays, CXR and skull X-ray (for jaw lesions, osteomas, supernumerary teeth).
- Colonoscopy with biopsies: investigation of choice for diagnosis.
- Upper gastrointestinal endoscopy: for evaluation of gastric and duodenal polyps.
- Genetic testing: for the APC gene and its mutation. Prenatal testing is possible if a disease-causing mutation is identified in an affected family member.

Differential diagnosis

The most important differential diagnosis to consider is colorectal cancer.

Other hereditary polyposis syndromes include:

- Adenomatous polyposis syndromes - eg, Turcot's syndrome.
- Hamartomatous polyposis syndromes - eg, Peutz-Jeghers' syndrome, juvenile polyposis and Cowden's disease.

Peutz-Jeghers syndrome

Peutz-Jeghers syndrome is an autosomal dominant disorder (with high penetrance) characterised by mucosal pigmentation of the lips and gums with multiple intestinal hamartomatous polyps.[7]

- There is an associated marked increased risk of certain malignancies, especially gastro-oesophageal, small bowel, colorectal and pancreatic. There is also risk of ductal breast cancer, thyroid, lung, uterine, Sertoli cell testicular tumours or ovarian sex cord tumours.[8]
- The estimate of the prevalence of Peutz-Jeghers syndrome is about 1 in 50,000.[9]
- In up to two thirds of cases, mutations can be identified in the serine/threonine kinase gene STK11(LKB1) on chromosome 19 (19p13.3).[10]

Symptoms

- Family history: asymptomatic but requesting investigation/counselling.
- Deeply pigmented lesions on the lips (cross the vermillion border) and buccal mucosa. These may also be present on the hands and feet (particularly the palms and the soles) and around the anus and genitalia. These lesions may be most prominent in infancy and fade after puberty.
- Repeated bouts of abdominal pain in a young patient (due to obstruction or intussusception).

- Unexplained intestinal bleeding in a young patient or iron-deficiency anaemia.
- May also present with rectal prolapse, precocious puberty, or with nasal, bronchial, biliary tract, uterine or bladder polyps.

Management

- If Peutz-Jeghers syndrome is suspected on the basis of clinical criteria, the patient should be referred to a regional genetics centre for formal counselling and mutation analysis of the relevant gene.[9]
- Surgical excision of lesions may be required:
- Endoscopic polypectomy for diagnosis and control of symptoms.
- Polypectomy using double balloon enteroscopy may prevent the need for repeated urgent operations and small bowel resection that leads to short bowel syndrome.[11]
- Colorectal surveillance: large bowel surveillance is recommended two-yearly from the age of 25 years. The intervention should visualise the whole colon and so colonoscopy is the preferred mode of surveillance.[9]
- Members of the family of an affected family where a causative gene has been identified should be referred for gene counselling and predictive gene testing. Where they test negative, there is no indication for their continued surveillance.[12]

Diseases associated with familial polyposis coli

The majority of patients have one or more extra-colonic features.[5] Extra-colonic features include:[1]

- Malignant tumours:
    - Duodenal.
    - Pancreatic.
    - Thyroid.
    - Brain (medulloblastoma).
    - Hepatoblastoma.
- Nasopharyngeal angiofibromas.
- Osteomas.
- Radiopaque jaw lesions.
- Supernumerary teeth.
- Lipomas, fibromas, epidermoid cysts.
- Desmoid tumours.
- Gastric adenomas.
- Duodenal, jejunal and ileal adenomas.

Familial polyposis treatment

Screening by flexible sigmoidoscopy with biopsy of polyps for histological diagnosis confirms the condition and allows surgery before the age of 20. See also the separate Screening for the Early Detection of Colorectal Cancer article. Non-surgical treatments that have been used include:

- Aspirin and celecoxib may reduce recurrence of adenomas and the incidence of advanced adenomas in individuals with an increased risk of colorectal cancer.[13]
- Treatment with sulindac, tamoxifen, or a combination of both, has been used for people with familial polyposis and desmoid tumours.[14]

Surgical

The types of surgery are:[6]

- Proctocolectomy with ileostomy.
- Total colectomy with ileo-rectal anastomosis.
- Restorative proctocolectomy with ileo pouch anal anastomosis - the main surgical treatment for patients with familial adenomatous polyposis.[15]

Complications

Complications secondary to polyps lining the colon include:

- Gastrointestinal haemorrhage.
- Gastrointestinal obstruction.
- Malignant change (if prophylactic colectomy is not performed). The risk of colorectal cancer increases with the number of polyps:
    - The mean age of colon cancer diagnosis in untreated individuals is 35-40 years.[16]
    - Patients with more than 1,000 polyps have been proven to have 2.3 times the cancer risk compared to patients with fewer than 1,000 polyps.

Screening for colorectal cancer

The British Society of Gastroenterology recommends:[17]

- High-risk criteria for future colorectal cancer comprise either:
    - Two or more premalignant polyps including at least one advanced colorectal polyp (defined as a serrated polyp of at least 10 mm in size or containing any grade of dysplasia, or an adenoma of at least 10 mm in size or containing high-grade dysplasia); or
    - Five or more premalignant polyps.
- People with high-risk findings on index colonoscopy who are under the age of 75 years should have a surveillance colonoscopy performed after an interval of three years. Surveillance should only be performed in people whose life-expectancy is greater than 10 years, and in general not in people older than about 75 years.

- People with no high-risk findings on index colonoscopy should not undergo colonoscopic surveillance, but should be strongly encouraged to participate in their national bowel screening programme when invited (see Further Reading below for link).
- People with premalignant polyps but no high-risk findings on index colonoscopy, who are more than 10 years younger than the national bowel screening programme lower age-limit, should be considered for a surveillance colonoscopy performed after an interval of 5 or 10 years, individualised to their age and other risk factors.
- Patients who have undergone a potentially curative colorectal cancer resection should have a clearance colonoscopy within a year of their diagnosis. Once a clearance colonoscopy has been performed in the postoperative period in patients who have had a colorectal cancer resection, their next surveillance should be performed after an interval of three years. The need for further surveillance should then be determined in accordance with the post-polypectomy high-risk criteria.
- The need for ongoing colonoscopic surveillance should be determined by the colonoscopic findings at each surveillance procedure, using the same high-risk criteria to stratify risk. People with high-risk findings on a surveillance colonoscopy should undergo a further surveillance colonoscopy at an interval of three years. People with no high-risk findings on a surveillance colonoscopy should cease colonoscopic surveillance, but should participate in the national bowel screening programme when invited.
- When colonic surveillance is required after previous polypectomy, computed tomography colonography (CTC) is an acceptable alternative if colonoscopy is incomplete or not possible due to the patient's clinical condition.
- When colonic surveillance is required after curative-intent resection of colorectal cancer, CTC should only be used for individuals in whom colonoscopy is contra-indicated or not possible due to the patient's clinical condition.
- Faecal immunochemical testing for surveillance after resection of premalignant colorectal polyps is not recommended, as there is insufficient evidence.

Prognosis[18]

Colorectal cancers generally derive from once benign dysplastic colorectal polyps. However, colorectal polyps are very common, affecting the majority of adults according to recent endoscopic screening data. The vast majority of colorectal polyps behave in a benign fashion and never develop into cancer.

However, colorectal cancer will develop in all individuals with familal polyposis unless prophylactic colectomy is performed. After total colectomy with ileo-rectal anastomosis, the recurrence rate is 30% after 20 years and 45% after 30 years.

Prevention

Screening family members by flexible sigmoidoscopy confirms or eliminates the diagnosis.

Further reading

- Colonoscopic surveillance for prevention of colorectal cancer in people with ulcerative colitis, Crohn's disease or adenomas; NICE Clinical Guideline (March 2011 - last updated September 2022)
- NHS Bowel Cancer Screening Programme
- Combined endoscopic and laparoscopic removal of colonic polyps; NICE Interventional procedures guidance, September 2014

Authored by:	Peer Reviewed by: Dr Hayley Willacy, FRCGP	
Originally Published: 20/11/2023	Next review date: 17/01/2023	Document ID: doc_2134

View this article online at: patient.in/doctor/bowel-colonic-polyps-pro

Discuss Bowel (colonic) polyps and find more trusted resources at Patient.