CONDITION

Familial adenomatous polyposis

Familial adenomatous polyposis is an inherited condition that causes many bowel polyps and a high colorectal cancer risk.

WHAT IT IS
Inherited condition causing many bowel polyps

BODY SYSTEM
Digestive system

MAIN SYMPTOMS
Rectal bleeding, diarrhoea, abdominal pain, unexplained anaemia

WHAT CAUSES IT
Inherited APC gene change

HOW IT IS DIAGNOSED
Colonoscopy, tissue examination, and APC genetic testing

MAIN TREATMENT
Endoscopic surveillance and preventive colorectal surgery

CAN IT BE CURED?
No, inherited cancer risk remains lifelong

IS IT AN EMERGENCY?
Sometimes, with bowel blockage or heavy bleeding

LONG-TERM OUTLOOK
Cancer risk greatly reduced with lifelong specialist care

Overview

Familial adenomatous polyposis, or FAP, is an inherited condition that causes many abnormal growths called polyps to develop in the colon and rectum. These are the final parts of the bowel.

Most polyps are adenomas. An adenoma is not cancer, but it can gradually become cancer. In classic FAP, hundreds or thousands of adenomas may develop, often beginning during the teenage years.

Without effective monitoring and treatment, colorectal cancer is expected to develop in nearly everyone with classic FAP, often at a younger age than usual. Timely surgery can greatly reduce this risk.

A milder form, called attenuated FAP, usually causes fewer polyps. They tend to appear later, but the risk of colorectal cancer is still high.

FAP can also affect other parts of the body. Polyps may form in the stomach and duodenum, the first part of the small bowel. Some people develop desmoid tumours, thyroid growths, bone changes, or other less common features. For this reason, follow-up continues even after colorectal surgery.

Symptoms

FAP often causes no symptoms at first. Polyps may be found through planned screening because the condition is known in the family.

When symptoms occur, they can include:

Anaemia means there are not enough healthy red blood cells to carry oxygen. Slow bleeding from polyps can cause iron-deficiency anaemia.

Symptoms do not show how many polyps are present or whether cancer has developed. Someone with many polyps may feel well, while similar symptoms can also come from common conditions unrelated to FAP. Testing is needed to understand the cause.

- Blood or mucus in the stool
- Diarrhoea or a change in bowel habits
- Abdominal pain or cramping
- Tiredness, weakness, or breathlessness from anaemia
- Unexplained weight loss

Causes and risk factors

FAP is usually caused by a harmful change in the APC gene. This gene normally helps control how cells grow. When it does not work properly, polyps can develop more easily.

The condition follows an autosomal dominant inheritance pattern. This means a person with an APC gene change has a 1 in 2 chance of passing it to each child. The chance is the same for every pregnancy.

Some people with FAP have no known family history. The genetic change may have started in them rather than being inherited from a parent. This is called a new, or de novo, genetic change. Family history alone therefore cannot rule out FAP.

Different APC changes can lead to different patterns. Some are linked with classic FAP, while others are more likely to cause attenuated FAP. Even within one family, the number of polyps and other features may not be exactly the same.

Other inherited conditions can also cause multiple bowel polyps. Genetic testing helps distinguish FAP from conditions involving genes such as MUTYH .

Diagnosis

A colonoscopy is a central test for FAP. A thin, flexible camera is passed through the rectum to examine the colon. Polyps can be counted, measured, removed, or sampled.

Removed tissue is examined under a microscope. This confirms whether a polyp is an adenoma and checks for cancerous changes.

A blood or saliva test can look for a harmful APC gene change. Finding one can confirm the genetic cause and allow close relatives to have targeted testing for the same change.

Genetic testing does not identify every case. FAP may sometimes be diagnosed from the number and pattern of adenomas, the person's age, and their family history, even when testing does not find an APC change. Broader genetic testing may be considered when another inherited polyposis condition is possible.

If FAP is confirmed, assessment may also include an upper endoscopy. This uses a flexible camera to examine the stomach and duodenum. Other tests are selected according to age, symptoms, family history, and the specific APC change.

Children in a family with a known APC change may be offered predictive genetic testing because bowel surveillance often needs to begin before adulthood. Genetic counselling can help families understand what the result may mean.

Treatment and management

Management aims to prevent cancer, find concerning changes early, and reduce the effects of polyps and other FAP-related growths.

Regular colonoscopy is used to monitor the number, size, and microscopic features of colorectal polyps. Small polyps may be removed during the procedure, but endoscopy alone usually cannot control the very large number found in classic FAP.

Most people with classic FAP eventually need surgery to remove the colon, sometimes together with the rectum. The timing is planned individually. Important factors include the

number and size of polyps, whether they can be controlled endoscopically, symptoms, biopsy findings, and personal circumstances.

The main operations include:

An ileostomy brings the end of the small bowel through an opening in the abdomen so waste can empty into a bag. An internal pouch is made from small bowel and connected to the anus.

Each operation has different effects on bowel frequency, fertility, continence, and the need for future procedures. The choice is made with a specialist colorectal team.

Surgery greatly reduces colorectal cancer risk, but it does not remove the inherited condition. Polyps can still develop in a remaining rectum, an internal pouch, the stomach, or the duodenum. Lifelong endoscopic surveillance remains important.

Certain medicines, including some non-steroidal anti-inflammatory medicines, can reduce colorectal polyp numbers in selected people. They are specialist treatments and do not replace surgery or surveillance.

Desmoid tumours may be watched with scans or treated with medicines, surgery, or other approaches. Management is individual because some remain stable while others grow or involve nearby organs.

- Removing the colon and joining the small bowel to the rectum
- Removing the colon and rectum and creating an internal pouch
- Removing the colon and rectum and creating a permanent ileostomy

Complications

The main complication is colorectal cancer. The risk becomes very high when classic FAP is not treated.

Other possible complications include:

Desmoid tumours are not cancer because they do not spread to distant organs. However, they can grow into nearby tissues and sometimes cause serious problems. They may appear after abdominal surgery, although surgery is not their only cause.

Some children with an APC gene change have an increased risk of hepatoblastoma, a rare liver cancer of early childhood. Families should receive advice based on the child's age and genetic findings rather than arranging tests without specialist guidance.

- Cancer in the duodenum or around the opening of the bile and pancreatic ducts

- Stomach polyps and, less commonly, stomach cancer
- Desmoid tumours, which can press on the bowel or other structures
- Thyroid cancer
- Bleeding, anaemia, or bowel blockage
- Polyps or cancer in a remaining rectum or surgically created pouch

Prevention

The inherited APC gene change cannot currently be prevented through diet, exercise, or lifestyle choices. A person does not cause FAP by anything they have done.

The complications of FAP can often be prevented or reduced. Genetic testing can identify relatives who need surveillance, while regular endoscopy and timely colorectal surgery can prevent many cancers.

Not smoking, staying active, and eating a balanced diet support general health, but they do not replace FAP-specific monitoring or surgery.

Living with it and what to expect

FAP requires lifelong follow-up. Before surgery, appointments often focus on colonoscopy, upper endoscopy, genetic care, and planning the timing and type of operation. After surgery, surveillance continues for any remaining rectum or pouch and for the stomach and duodenum.

Bowel habits usually change after colorectal surgery. Stools may be more frequent or looser, particularly during recovery. Many people adapt over time, although the long-term result differs by operation and individual.

People with FAP commonly continue education, employment, travel, relationships, and family life. Endoscopy, surgery, bowel symptoms, and concern about relatives can still affect daily routines and emotional wellbeing. Stoma nurses, dietitians, genetic counsellors, and psychological support can help with these practical and emotional changes.

Follow-up plans may change over the years. More frequent examinations may be needed if polyps become larger, more numerous, or harder to remove. New symptoms may also lead to imaging or earlier endoscopy.

Family planning can involve difficult choices. Genetic counselling can explain natural conception, testing during pregnancy, and assisted reproduction with testing of embryos.

These are personal options, not requirements.

When to get medical help

Arrange a routine appointment if you have a family history of FAP, multiple bowel polyps, or a relative with a harmful APC gene change. Genetic assessment may be appropriate even if you feel well.

If you already have FAP, contact your care team promptly about new rectal bleeding, persistent changes in bowel habits, unexplained weight loss, worsening abdominal discomfort, a neck lump, or a new abdominal mass. These symptoms need assessment but do not automatically mean cancer.

Seek urgent medical help for:

Call emergency services for fainting, confusion, severe breathlessness, or signs of shock such as cold, clammy skin after significant bleeding.

Routine surveillance should continue even when you feel well. It is designed to find changes before they cause symptoms.

- Severe or rapidly worsening abdominal pain
- Repeated vomiting with a swollen abdomen
- Inability to pass stool or gas
- Heavy or continuing rectal bleeding
- Black, tar-like stools with weakness or dizziness

Common questions

Is FAP cancer?

No. FAP is an inherited condition that causes polyps and greatly increases cancer risk. Most adenomas are not cancer when they first form. The concern is that some can become cancer over time.

Will I definitely get bowel cancer if I have FAP?

Untreated classic FAP leads to an extremely high lifetime risk of colorectal cancer. Surveillance and timely surgery can greatly reduce that risk. Attenuated FAP also carries a high risk, although cancer often develops later.

Can I have FAP without anyone else in my family having it?

Yes. An APC gene change can begin for the first time in one person. A small family, limited knowledge of relatives' health, or relatives dying young from other causes can also hide an inherited pattern.

Does everyone with FAP need their colon removed?

Most people with classic FAP eventually need colorectal surgery because endoscopy cannot safely control hundreds or thousands of polyps. Some people with attenuated FAP can be managed endoscopically for longer. The decision is based on the actual polyp burden and biopsy findings.

Can I pass FAP to my children?

A person with an APC gene change has a 1 in 2 chance of passing it to each child. A child who does not inherit that family change cannot pass it on. Genetic counselling can explain testing and reproductive options.

What is the difference between FAP and attenuated FAP?

Classic FAP usually causes hundreds or thousands of colorectal adenomas beginning relatively early. Attenuated FAP generally causes fewer polyps and tends to lead to cancer later. Both require lifelong specialist follow-up.

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