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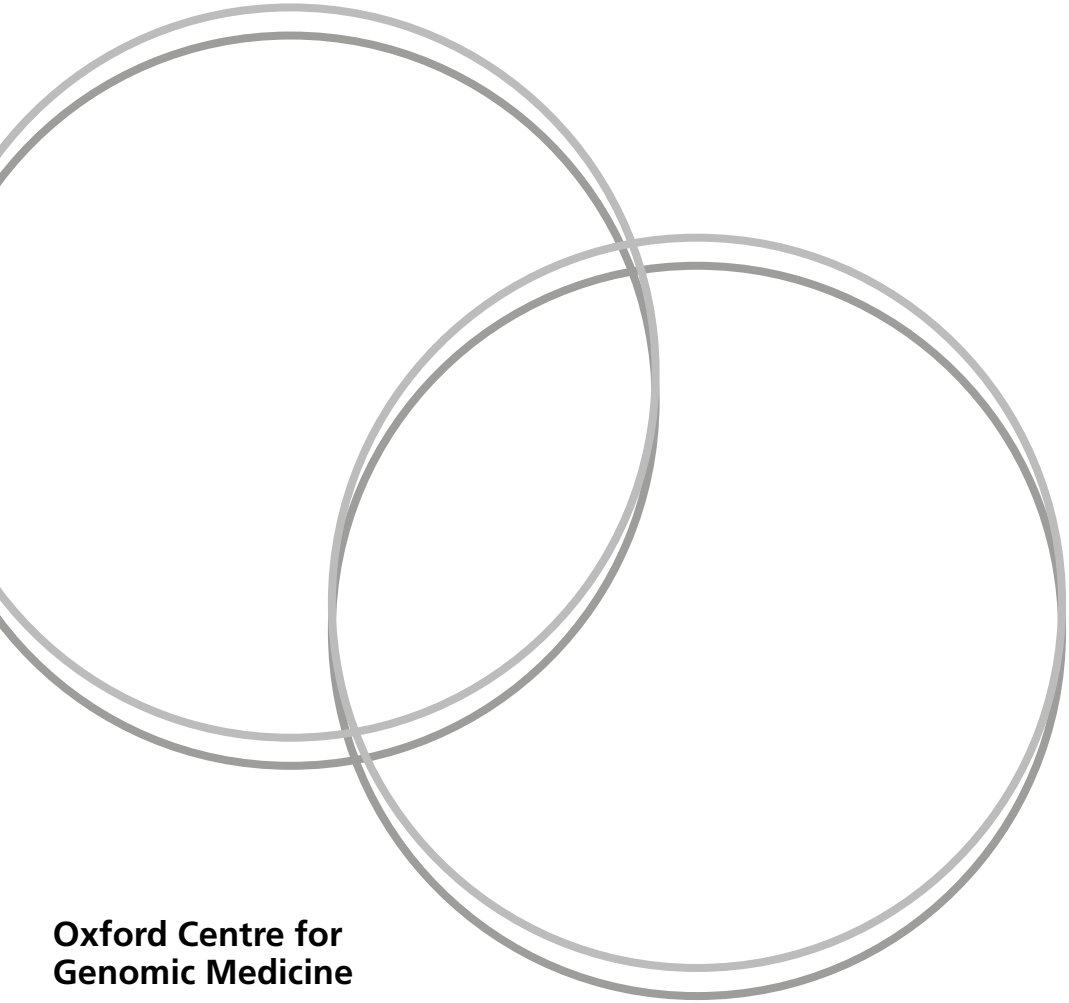
Familial Adenomatous Polyposis (FAP)

Information for patients and families



Oxford Centre for Genomic Medicine

What is FAP?

FAP is an inherited condition which causes people to be more at risk of developing bowel polyps and cancers.

What is a polyp?

A polyp is a small non-cancerous growth. Polyps usually occur in the colon (large bowel). It is normal for an adult to develop 1 or 2 polyps as they get older but it is unusual to have lots of polyps. When lots of polyps occur, we call this Polyposis.

What happens in FAP?

In FAP there is a tendency to develop large numbers of a certain type of polyp. These polyps are called adenomatous polyps or adenomas. They usually develop in the colon. They can also occur in the stomach or small bowel. Other names for FAP are hereditary polyposis of the colon, familial polyposis, and Gardner syndrome. In FAP polyps start to develop throughout the colon, usually in teenagers or young adults. Adults may have hundreds to thousands of polyps. The major concern is that the polyps will become cancerous.

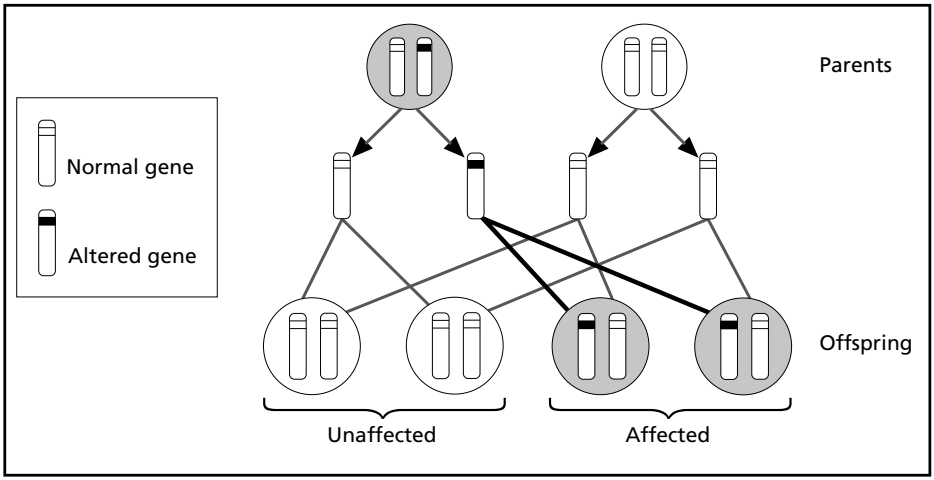
What is Attenuated FAP (AFAP)?

Some families have Attenuated FAP (AFAP). This is a mild form of FAP. Individuals with AFAP develop fewer polyps at a later age than those with typical FAP. People with AFAP may develop colon cancer from these polyps at a later age than someone with FAP but also still have a very high risk of developing colon cancer during their lifetime. If AFAP is suspected within a family it is important that family members are offered bowel screening, as discussed later in this leaflet. You should discuss with your geneticist when screening should begin.

How does FAP run in families?

Genes are instructions which tell our bodies how to work. We each have about 30,000 genes. All our genes come in pairs as we get one copy from our Mother and one from our Father. Each gene has a specific job.

In FAP, part of a gene called *APC* is missing or altered, so it cannot do its job properly. Lots of polyps develop because of the altered copy of the gene. If someone has FAP it means that they have one working copy and one altered copy of the *APC* gene. Each time they have a child, there is a fifty-fifty chance that the child will inherit the altered copy, and a fifty-fifty chance the child will inherit the working copy. This is shown in the diagram below.



What about genetic testing?

The gene involved in FAP (called *APC*) is like a very long instruction. A spelling mistake or alteration in any part of this instruction can cause FAP. Each family with FAP will have a different alteration. We can look in a blood sample from someone who has FAP to try to find the alteration in a family.

What happens if you don't find an alteration in my family?

In order to offer a genetic test to other relatives we first need to find the gene alteration responsible for causing FAP in a family. If we do not find a gene alteration, we will not be able to offer a genetic test to other relatives but they will still need to undergo bowel screening. The starting age of bowel checks and the frequency they are performed will depend on how many polyps other relatives with FAP have had, and how old they were when they started to develop polyps. We will discuss the best screening plan for your family with you.

Most people with FAP develop polyps by the age of 40. Therefore, people who are older than this who have not developed any polyps are unlikely to have FAP and can stop bowel screening.

What happens if you do find a gene alteration in my family?

If we do find an alteration, we can test other family members to see if they have inherited it. We recommend that children have a gene test around the time of puberty to see if they have inherited the altered gene.

If they have not inherited the altered gene, they do not need bowel checks. They will not be able to pass the altered gene on to their children in the future.

If your children have inherited the altered gene it is important that they have bowel checks every year, starting from puberty.

How do we know who might have FAP?

Your family may have FAP if:

- Several family members have had colon cancer or polyps.
- One of your relatives has had many polyps.
- A relative has had colon cancer at a young age with polyps.

FAP can have other effects on the body as well as causing polyps. Some people get skin cysts, some get harmless bumps on their bones and some get freckle-like spots on the inside lining of the eye, which we call CHRPEs. These do not affect vision in any way.

Some people develop benign (non-cancerous) growths in their abdomen called desmoid tumours. These tumours sometimes need to be removed if they cause symptoms but they do not become cancerous. There is no need to have regular checks for desmoids but if you have any lumps or unexplained symptoms, you should discuss them with your GP.

What bowel checks are needed for families with FAP?

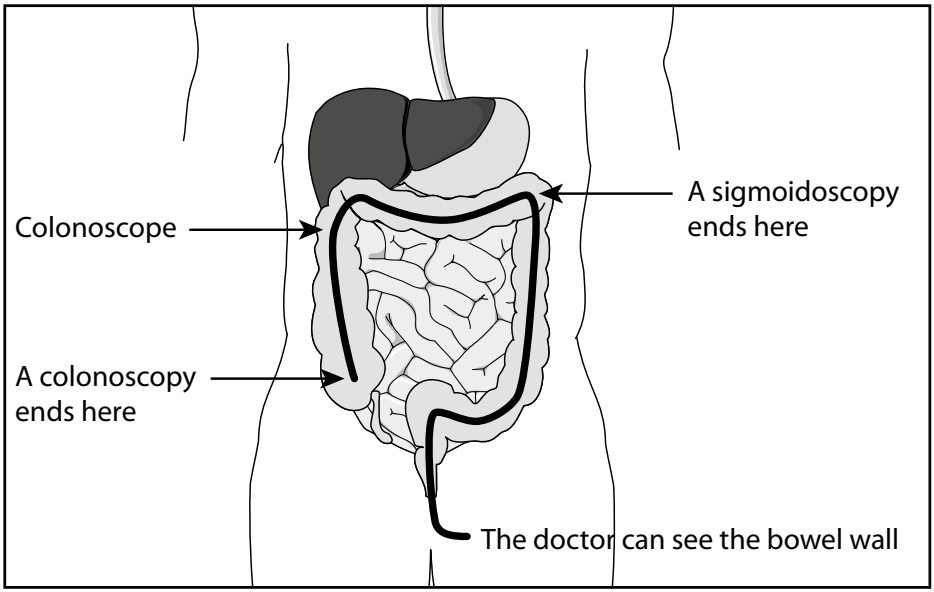
Most people who have FAP begin to develop polyps in their early teens. People at risk of FAP should start bowel checks around puberty. These should be repeated every year. It is important to remember that it is not safe to simply wait for symptoms to develop. It is vital that children at risk of FAP have regular bowel checks even if they do not have symptoms.

Polyps can sometimes occur in other areas of the body, although not as often as in the bowel. We will discuss with you whether any other checks are appropriate in your family.

What do bowel checks involve?

A short tube holding a thin flexible camera is passed through the back passage into the lower part of the bowel. This examination is called a sigmoidoscopy. A longer, flexible tube can be used to examine more of the bowel. This is called a colonoscopy. Doctors may use either method to screen the bowel. It is important to prepare the bowel by emptying it before the test. The hospital will give you more details about this. It usually involves taking a liquid medicine (a laxative) the day before the check. You can usually go home from hospital on the same day as your screening.

How the bowel is examined by colonoscopy and sigmoidoscopy



What happens if polyps are found?

If polyps are found, the doctor will take a biopsy (tiny sample), to be looked at under a microscope. If the polyps are harmless, it may be alright to carry on with regular checks. However, if they find a large number of polyps, most doctors will offer the choice of an operation. Many people choose to have an operation as soon as polyps are found, rather than wait until there are large polyps.

As more polyps will continue to develop after others have been removed, it becomes too difficult to remove all the polyps. Most people with FAP eventually opt to have surgery to remove the large bowel completely.

What types of operation are available for FAP?

An ileorectal anastomosis (IRA) involves removing the large bowel and attaching the small bowel to the rectum. The rectum may develop polyps so this must be examined at least once a year to prevent a cancer starting there.

A pan-proctocolectomy is done when the rectum has to be removed. In this operation the end of the small bowel is bought out to the skin and the body's waste material is collected in a disposable bag (an ileostomy).

In a pouch operation the colon is removed along with the lining of the rectum. A pouch is then made from the end of the small bowel and the rectum. No checks have to be made on the rectum as there is no lining to grow polyps.

Both the IRA and the pouch operation would allow you to go to the toilet normally but sometimes you may go more frequently than before.

Is there a cure for FAP?

There is lots of research looking at FAP which may help us to develop different treatments in the future. There is no way of correcting or replacing the altered gene at the moment. Doctors are looking at non-surgical ways to stop polyps growing, such as drug treatment. These may offer a way to treat FAP without surgery in the future. At the moment, the only way to reduce the risk of cancer in FAP is with an operation.

Who should I contact for further advice?

The first person you should ask if you have any health problems is your GP. He/she may not be very familiar with this problem, so you should take along this leaflet. If anyone in your family has FAP, you could ask their surgeon for advice. Every region in the UK has a genetics service who can be contacted for advice via your GP or directly. Individuals who live in the Oxfordshire region are welcome to contact us for further information.

Explanation of unfamiliar words

Familial	Something which runs in the family.
Polyp	A non-cancerous growth on the bowel wall.
Adenoma (Adenomatous)	There are different kinds of polyp: this describes how polyps in FAP look under the microscope.
Large bowel	The end section of the intestine made up of the colon and rectum.
Gene	One of the instructions which control the working of the body.
CHRPE	(Congenital hypertrophy of the retinal pigment epithelium.) Harmless black marks on the back of the eye.
Sigmoidoscopy	A short thin tube, about the thickness of your little finger, containing a camera is passed into the rectum and the last part of the colon to look for polyps. A light sedation can be given.
Colonoscopy	Like sigmoidoscopy, but the whole colon is checked.
APC	The name scientists give the FAP gene. It stands for adenomatous polyposis coli.

If you need more advice please contact:

Oxford Centre for Genomic Medicine

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Further information

If you would like an interpreter, please speak to the department where you are being seen.

Please also tell them if you would like this information in another format, such as:

- Easy Read
- large print
- braille
- audio
- electronic
- another language.

We have tried to make the information in this leaflet meet your needs. If it does not meet your individual needs or situation, please speak to your healthcare team. They are happy to help.

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