

## Explanation of unfamiliar words

|                       |                                                                                                                   |
|-----------------------|-------------------------------------------------------------------------------------------------------------------|
| Familial              | Something which runs in families.                                                                                 |
| 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 tube with a light at the end is passed into the rectum and the last part of the colon to look for polyps. |
| Colonoscopy           | Like sigmoidoscopy, but the whole colon is checked.                                                               |
| APC                   | The name scientists give the FAP gene. It stands for adenomatous polyposis coli.                                  |

## Further Information

<http://www.cancerresearchuk.org/>

<http://www.macmillan.org.uk/Home.aspx>

*Birmingham Women's NHS Foundation Trust is not responsible for the content of third-party information and does not endorse any product, view or process or opinion from such sources.*

Reference Number: CG5/FAP

Author: P Leonard, K Ong,

Date created Feb 2010/Review Date: Feb 2013

**Birmingham Women's NHS Foundation Trust**

**Edgbaston, Birmingham , B15 2TG**

**Telephone: 0121 472 1377**

**Fax : 0121 627 2602**

# Birmingham Women's



NHS Foundation Trust

## Familial Adenomatous Polyposis

### *Information for patients and families*

**If you need more advice please contact:**

West Midlands Family Cancer Service

Clinical Genetics Unit

Birmingham Women's NHS Foundation Trust

Mindelsohn Way

Edgbaston,

Birmingham

B15 2TG

Telephone: 0121 607 4757

Fax: 0121 623 6949

Email: [wmfacs@bwhct.nhs.uk](mailto:wmfacs@bwhct.nhs.uk)

## What is FAP?

FAP is an inherited condition which causes people to be predisposed to 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. Although people with AFAP tend to develop colon cancer at a later age, they still have a very high lifetime risk of colon cancer. If AFAP is suspected within a family, it is important that family members are screened with colonoscopy. As AFAP is variable you should discuss when screening should begin with your geneticist.

In a *pouch* operation the lining of the rectum and the colon are removed. A pouch is then made from the end of the small bowel. No checks have to be made on the rectum as there is no lining to grow polyps. Both these operations would allow you to go to the toilet normally, but sometimes this can be many times a day.

A *pan-procto colectomy* is done when the rectum has to be removed. The end of the small bowel is brought to the surface and the waste material is collected in a disposable bag (an ileostomy).

## 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. Scientists are working on ways to put a correct copy of the FAP gene into the bowel wall. This is called "gene therapy".

Doctors are also 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.

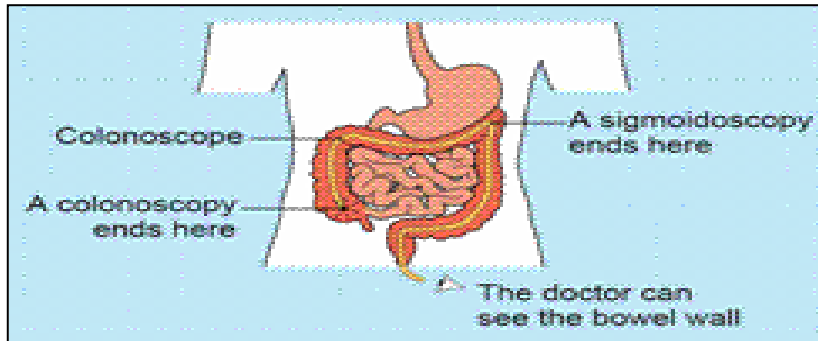
## Who should I contact for further advice?

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 help via your GP or directly for advice.

Individuals who live in the West Midlands are welcome to contact us for further information.

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. Most people with FAP will eventually have surgery to remove the large bowel.

### **What types of operation are available for FAP?**

An *ileo-rectal 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.

### **How do we know who has 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.

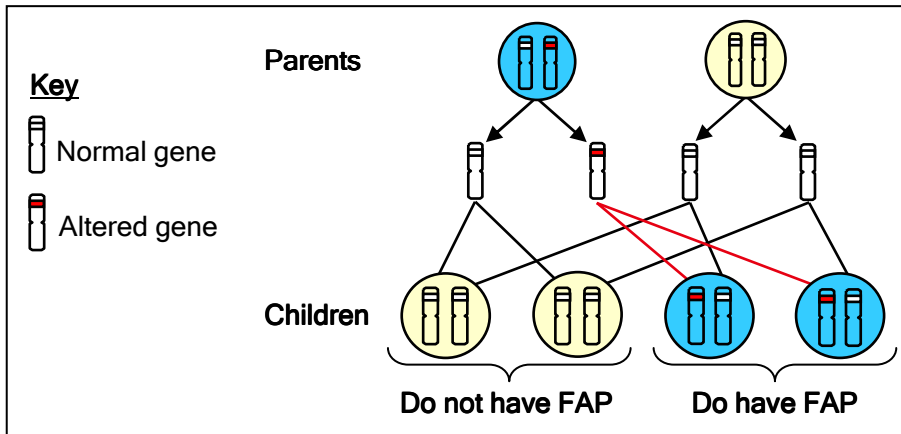
FAP can have other effects on the body as well as causing polyps. Some people get skin cysts, some get harmless bumps on their bones and some get freckle-like spots on the inside lining of the eye, which we call CHRPEs. These do not affect vision in any way.

Some people develop benign 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.

### **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 Mum and one from our Dad. 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 overleaf.



### 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?

If we don't find a gene alteration, we won't be able to offer a genetic test to other relatives but they will still need bowel checks. The frequency and starting age of bowel checks depends on your family history. 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 checks.

### What happens if you do find an 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 age of 10-12 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 they have inherited the altered gene* it is important that they have bowel checks every year starting from the age of 10-13. All the children in the west midlands with FAP have their checks at a specialist children's unit in Birmingham.

### 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 between 10 and 13 years of age. 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 can be 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.