

Familial adenomatous polyposis (FAP)

Information for patients



This booklet has been written for people attending the clinical genetics service with a personal or family history of familial adenomatous polyposis (FAP).

What is familial adenomatous polyposis (FAP)?

FAP is an inherited condition that runs in families, hence the term familial. The rest of the name comes from the fact that little lumps called polyps grow in large numbers on the lining of the bowel.

What is a polyp?

There are several different types of polyps that can grow on the lining of the bowel (some are common with increasing age). The type of polyps seen in FAP are called adenomas, hence the term 'adenomatous'. The polyps themselves are not cancerous, but may become so if left to grow. People who have inherited FAP will eventually develop cancer if they do not receive screening and treatment.

A diagnosis of FAP is made when large numbers of adenomas are found in the large bowel (called the colon and rectum), which is the end of the long food pipe of the body. A diagnosis of FAP can also be made using a genetic test.

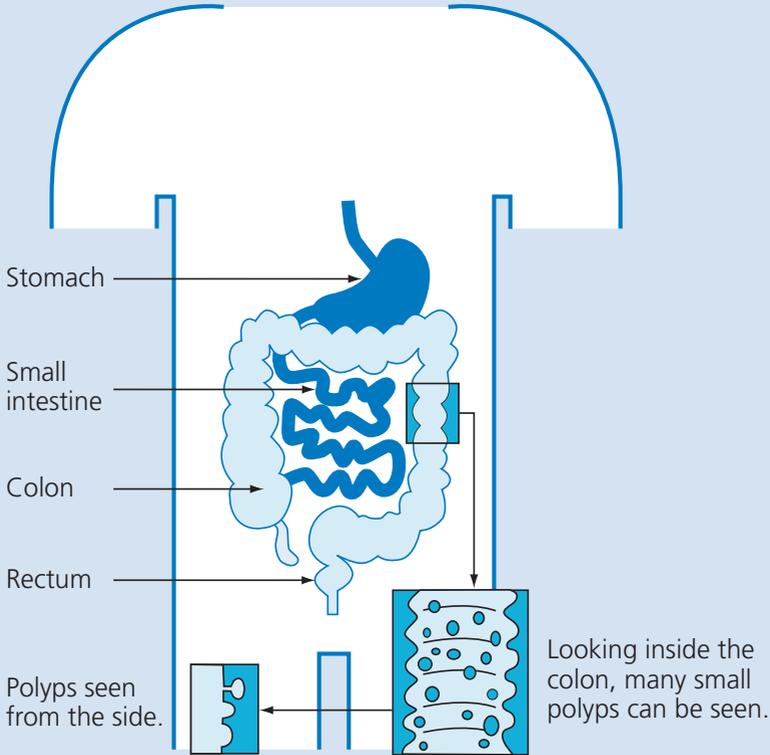
How do we look for polyps?

The lining of the bowel can be examined through a procedure called a colonoscopy, in which a long flexible tube (colonoscope) is passed through the anus into the large bowel. The colonoscope has a bright light on one end and a camera on the other so that the surgeon can get a clear view of the lining of the bowel.

This procedure is usually carried out as a day case so you will not need to stay in hospital. In order to see the lining of the bowel clearly it is necessary to prepare (empty) the bowel beforehand; this usually involves taking laxatives the day before.

We recommend that all people who are at risk of FAP are examined for bowel polyps once every year usually beginning between the age of 10 and 14 years old. Most people who have the condition develop polyps by age 30.

The large bowel (colon and rectum) showing polyps in the colon



After the test

If polyps are found at colonoscopy the surgeon will usually take a sample of these and the experts will look at them under the microscope. Depending on how the polyps look and the total number of polyps you have, the surgeon may recommend an operation to remove the large bowel (colon), or it may be reasonable to continue with screening. If one of the polyps gets bigger or looks like it could be turning into a cancer, the surgeon will recommend an operation.

This surgery may take place as early as 16 to 17 years of age, but the age varies quite a lot for different families.

Other symptoms

Polyps

Individuals with FAP may also develop polyps along other sections of the gut including the stomach and small intestine. Therefore, we also recommend that people with this condition should have a camera test of the upper part of the gut (gastroscopy) by the age of 30 years. This test is then repeated every 3 years.

Skin and bone cysts

Some people with FAP may develop skin lumps and cysts. They may also develop bone cysts which usually do not cause any clinical problems. These alone are not enough to make a diagnosis, but if they are found in children who have a family history of FAP, this suggests that the child has inherited the condition.

CHRPEs

Sometimes people with FAP get harmless dots at the back of the eye. These are called CHRPEs (this stands for congenital hypertrophy of the retinal pigment epithelium). These dots do not affect eyesight in any way. An eye doctor (ophthalmologist) can look at the back of the eye with a bright light to see if there is any evidence of CHRPEs.

A lot of people who do not have FAP have one or two CHRPEs, but if there are more than five or a large distinctive area, then this is an indication that a person may have the condition. However, not all people with FAP have CHRPEs.

Sometimes if these features, which develop outside the bowel, are present, the term Gardner's syndrome is used. There are other, less common features of FAP that develop in a small number of people, which we have not listed here. Your doctor can discuss these with you.

Causes

FAP is usually inherited, but occasionally a person may be the first member of their family to develop the condition.

Our bodies contain thousands of coded messages called genes which send instructions to our body about how to function; for example, what colour to make our eyes or how tall we will grow. There are also genes that are involved in repairing the damage to our bodies that occurs during normal daily life. If one of these genes is altered or faulty they will not function correctly and eventually cancer may result.

You may hear many different words used to describe a gene that is not working properly. A gene may be said to be faulty, altered or changed. The technical term is a mutation. This means that the instruction the gene sends to the body may be different, just as a spelling mistake may alter the meaning of the word.

How are these genes inherited?

We all have two copies of each of our genes; we get one copy from our mother, the other copy from our father. Some genes are so important that you need both copies working correctly to remain healthy. One faulty copy of these important genes can cause disease to develop.

When we have children, we pass just one of each copy on; the other comes from the other parent. If a person has a gene mutation, they will also have a normal copy. Therefore each of his or her children has a one in two (50%) chance that they will inherit the gene mutation. If a person has not inherited a mutation, then they cannot pass it on to their children.

Diagnosis

It's possible to look for the specific mutation in the gene which causes FAP. This can be done from a blood sample, because blood cells contain copies of all our genes. We start by looking at the genes of an affected person. If a mutation can be identified in the gene for FAP (called the APC gene) then other family members can be tested to see whether or not they have inherited the same mutation.

Because the gene mutation can be in many different places this initial testing can take several months. Once the family mutation has been found, testing of other individuals is quicker (usually a matter of weeks). This second stage is known as predictive testing.

If a person does not carry a familial mutation they can stop their bowel screening and they cannot pass the faulty gene on to their children.

This genetic test will find a gene mutation in 90 to 95% of people who clinically have FAP. If the mutation has not been found, then it may be possible to track the disease through the family using DNA markers. This will give a good indication of who is likely to carry the faulty gene within the family, but it is not quite as direct as a test for a specific gene mutation.

If a person does carry a familial mutation they should continue with yearly bowel screening until polyps are found. At this point the surgeon will usually recommend surgery, but this will usually have been discussed with you well before that stage.

Considerations

Some people experience strong emotions when they are told they have a gene that increases their risk of cancer. Anger, shock, anxiety about your health, worry or guilt about possibly passing the gene on to children are all normal reactions.

Some people may feel guilty if they have not inherited the gene mutation when other close family members have. Genetic testing in a family can affect other family members. They may need to be told they too are at increased risk, or unexpected information may be revealed. For example, someone may disclose that a family member is adopted.

Alternatives to genetic testing

You may choose not to have genetic testing, but you should have regular bowel screening as at-risk people who are screened regularly live longer than those who do not.

Treatment

The surgeon will discuss with you the main choices for surgery. Here is a brief outline of the most commonly performed operations.

1. Ileo-rectal anastomosis (IRA)

The simplest operation involves removing the colon and attaching the small intestine (ileum) to the rectum. This allows you to open your bowels normally, but this can sometimes be many times in one day.

The rectum, which remains in place, may develop polyps so this still needs to be screened regularly. In some patients a further operation may be needed several years later if many polyps grow in the rectum. This operation can often be done using keyhole surgery.

2. A pouch operation

This procedure involves two separate operations. The colon and rectum are removed and a portion of the small bowel is used to make a pouch that can store faeces (stools) and function like an artificial rectum. The first operation creates a temporary arrangement where the end of the small bowel is brought out through the abdominal wall (an ileostomy).

Once the new pouch has healed up fully, a second operation is needed to rejoin the pouch to the upper bowel so that you can go to the toilet in the usual way. Although a bigger procedure, this removes more bowel and therefore screening of the rectum is not required as it would be after an IRA.

3. Pan-proctocolectomy and ileostomy

This involves the removal of the colon and the rectum. The end of the small bowel is brought to the skin surface of the abdomen (called an ileostomy) and faeces are collected in a disposable bag. This operation means there is no large bowel left that is at risk from polyps.

All these procedures have advantages and disadvantages. You will need to discuss the options in detail with your surgeon to ensure that you choose the operation that is right for you when the time comes.

Glossary

Adenoma (Adenomatous)

A particular type of polyp that has the potential to become cancerous.

APC

The name scientists give the FAP gene. It stands for adenomatous polyposis coli.

CHRPE (congenital hypertrophy of retinal pigment epithelium)

Harmless black marks on the back of the eye.

Colectomy

An operation to remove the colon, leaving the rectum in place.

Colonoscopy

A long flexible tube is passed up the back passage to look at the inside of the bowel. If polyps are found they can be removed there and then.

Dominant inheritance

Occurs when there is a one in two (50%) chance of passing on a condition.

Familial

A condition which runs in families.

Gene

One of the chemical recipes (or coded messages) which control the working of the body.

Ileo-rectal anastomosis (IRA)

The small bowel is attached to the rectum when the colon has been removed.

Ileostomy

An opening in the abdominal wall for the passage of faeces.

Large bowel

The end section of the intestine or food pipe, made up of the colon and rectum.

Pan-proctocolectomy (PPC)

The colon and rectum are removed and the small bowel is brought to the surface of the abdomen.

Polyp

A non-cancerous lump on the bowel wall.

Pouch

A similar operation to IRA but the lining of the rectum is also removed and replaced by lining from the small bowel.

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.

The team involved in your care are:

Consultant:

Tel no:

Genetic counsellor:

Tel no:

Please contact us on 023 8120 6170 if you have any other questions.

Further information

If you have questions or need advice about any aspect of FAP, please contact us at:

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Version 4. Published May 2016. Due for review May 2019. GEN005.01