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Familial adenomatous polyposis (FAP)

Information for patients

We have written this booklet for people attending the clinical genetics service with a personal or family history of familial adenomatous polyposis (FAP). We hope this booklet will help to answer some of the questions you may have about FAP.

What is familial adenomatous polyposis (FAP)?

FAP is a rare condition that usually runs in families (this is what 'familial' stands for). The rest of the name comes from the fact that the condition causes little lumps (called polyps) to grow in large numbers on the lining of the bowel. Clinically, FAP is defined as more than 100 bowel polyps. If someone has multiple bowel polyps, but less than 100, this is called 'attenuated FAP'.

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 (this is what 'adenomatous' stands for). The adenomas themselves are not cancerous but may become so if left to grow. People with FAP will eventually develop bowel cancer if they do not receive screening and treatment for their condition.

A clinical diagnosis of FAP is made when large numbers of adenomas are found in the large bowel (called the colon and rectum). A diagnosis of FAP can also be made using a genetic test.

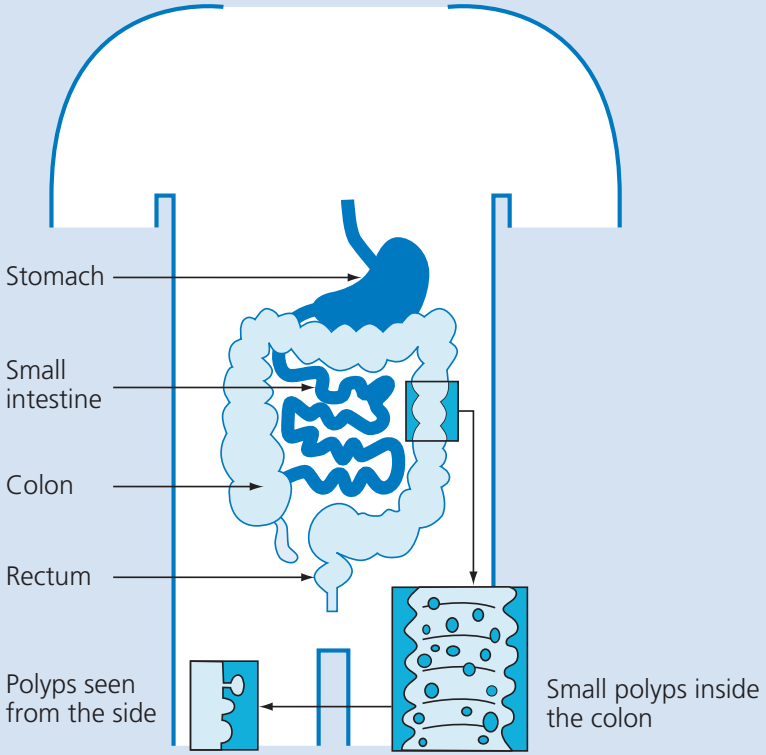
How do we look for polyps?

The lining of your bowel can be examined through a procedure called a colonoscopy. The procedure will involve a clinician passing a long flexible tube (called a colonoscope) into your large bowel through your back passage. The colonoscope has a bright light on one end and a camera on the other so that the clinician can get a clear view of the lining of your bowel.

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

We recommend that everyone who is at risk of FAP is examined for bowel polyps once a year, usually from the age of 10 years old. Most people who have the condition develop polyps by the time they are 30 years old.

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



What will happen after the procedure?

If polyps are found during your colonoscopy, the clinician will usually take a sample of these, so they can be looked at under a microscope. Depending on the size of the polyps, how they look and the total number of polyps you have, the clinician may recommend an operation to remove your large bowel (colon), or it may be possible for you to continue with screening for the time being.

The clinician may recommend having surgery from as early as 16 or 17 years of age. However, this age will vary for different families, so it may be some years later.

Are there any other symptoms of FAP?

Polyps

Individuals with FAP may also develop polyps along other sections of their gut, including their stomach and small intestine. Therefore, we also recommend that people with this condition have a camera test of the upper part of their gut (called a gastroscopy) by the time they are 30 years old. This test will then need to be repeated every three years.

Skin and bone cysts

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

CHRPEs

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

People who do not have FAP may 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 FAP. However, not everyone with FAP will have CHRPEs.

Desmoid tumours

Desmoid tumours are non-cancerous growths that occur in connective tissue. Some desmoid tumours are slow-growing and don't need immediate treatment. Others grow quickly and are treated with surgery, radiation therapy, chemotherapy or medication. Desmoid tumours are not considered to be cancerous because they don't spread to other parts of the body. However, they can be aggressive and grow into nearby structures and organs.

If any of these other symptoms of FAP are present, the term 'Gardner's syndrome' is sometimes used. There are also some 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.

What causes FAP?

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 everyday life. If one of these genes has a variant, it will not function correctly and eventually, it may result in cancer.

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

How are these genes inherited?

We all have two copies of each of our genes. We get one copy from our mother and 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 copy of each gene on. The other comes from the other parent. If a person has a gene variant, they will also have a second copy of the gene which is working normally. Therefore, each of their children has a one in two (50%) chance that they will inherit the gene variant. If a person has not inherited a variant, then they cannot pass it on to their children.

How is FAP diagnosed?

It is possible to look for the specific gene variant which causes FAP. This can be done from a blood sample. We start by looking at the genes of an affected person. If we identify a variant in the FAP gene (called the APC gene), then other family members can be tested to see whether they have inherited the same variant.

This genetic test will find a gene variant in 90 to 95% of people who clinically have FAP. If a person does carry a gene variant, they should continue with yearly bowel screening until polyps are found. At this point, the clinician will then usually recommend surgery.

Are there any alternatives to genetic testing?

You may choose not to have genetic testing, but you should still have regular bowel screening. Bowel screening involves having a colonoscopy every one to three years (depending on how many polyps are found). Screening usually starts between the ages of 10 and 14.

How is FAP treated?

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

Ileo-rectal anastomosis (IRA)

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

After this operation, you would still need to have your rectum screened regularly for polyps. You may also need a further operation several years later if many polyps grow in your rectum. This operation can often be done using keyhole surgery, which is minimally invasive.

A pouch operation

This operation involves removing your colon and rectum, and then using a portion of your small bowel to make a pouch that can store faeces (poo) and function like an artificial rectum. The operation is split into two separate operations.

The first operation creates a temporary arrangement where the end of your small bowel is brought out through your abdominal wall (an ileostomy).

Once the new pouch has healed fully, a second operation is needed to rejoin the pouch to your upper bowel so that you can go to the toilet in the usual way.

Although a more invasive procedure, you would not need to have your rectum screened after having it, as you would do after an IRA.

Pan-proctocolectomy and ileostomy

This operation involves removing your colon and rectum, and then bringing the end of your small bowel to the skin surface of your abdomen (called an ileostomy), so your faeces (poo) can be collected in a disposable bag. This operation means you have no large bowel left which is at risk from polyps.

All of these operations have advantages and disadvantages. You will need to discuss the options in detail with your clinician 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 anastamosis (IRA)
An operation to remove the colon and attach the small bowel to the rectum.

Ileostomy
An opening in the abdominal wall for the passage of faeces (poo).

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

Pan-proctocolectomy (PPC)
An operation to remove the colon and rectum, and bring the small bowel 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

Consultant: .....

Telephone: .....

Genetic counsellor: .....

Telephone: .....

Further information

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

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Notes

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For help preparing for your visit, arranging an interpreter or accessing the hospital, please visit **www.uhs.nhs.uk/additionalsupport**

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