Information Leaflet

NHS
Liverpool Women's
NHS Foundation Trust

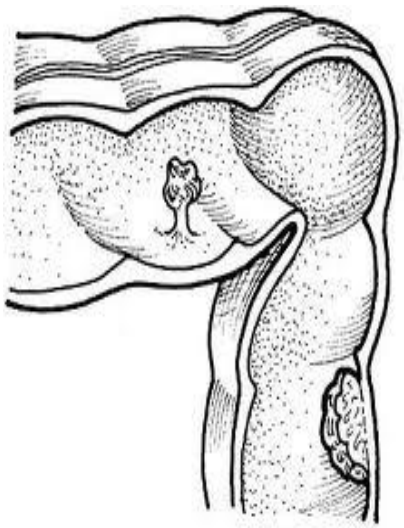
Familial Adenomatous Polyposis (FAP) in the family

What is Familial Adenomatous Polyposis (FAP)?

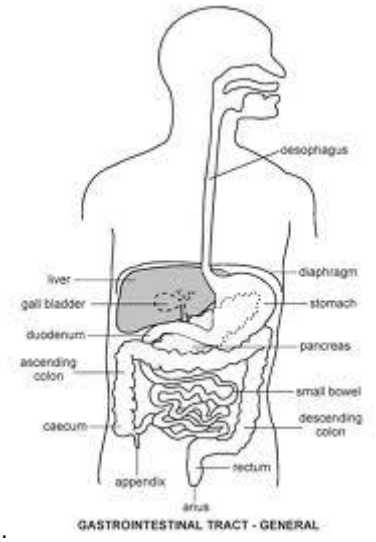
FAP is a condition which runs in families, hence the term **"familial"**. The rest of the name comes from the fact that little lumps called **polyps**, grow in large numbers on the lining of the bowel. The polyps themselves are not cancerous, but people who have FAP will eventually develop cancer if they do not receive treatment.

What is a polyp?

Polyps are non-cancerous lumps, or tumours, which grow on the surface of the bowel. There are many different kinds of polyp which grow in a number of places in the body, but the polyps we see in the bowel of people with FAP are called **adenomas**. Bowel polyps are found in many people without FAP, but usually just a few polyps are found. A diagnosis of FAP is made when large numbers of adenomas are found in the large bowel, which is the end of the long food pipe of the body which we call the colon and rectum. The following diagram shows where the colon and rectum are in the body, and what the polyps look like inside the bowel.



Laurel Cook Lhowe



GASTROINTESTINAL TRACT - GENERAL

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How is FAP inherited?

FAP is due to a fault in a gene that can be passed down families form a parent to their child. **Genes** are messages which control the working of the body and decide things like the colour of our eyes. Think of genes as recipes. The recipes allow the body to make chemical tools and building blocks. If part of the recipe is missing or has been copied incorrectly, that part of the body will not be able to do its job properly.

Most genes come in matching pairs, with a copy of each gene coming from the mother's egg and a copy from the father's sperm. We think that everyone has at least one gene which doesn't work, but usually the spare copy from the other parent is enough to get by on. Some genes seem to be so important that you need both copies working to remain healthy. One faulty copy of a gene of this type can cause disease, so we call this gene **dominant** because it shows itself despite the presence of a normal copy.

FAP is one of the more frequent dominant genetic problems, but it is still uncommon, affecting about one person in every 8,000.

If someone has FAP it means that they have a working copy and a faulty copy of the gene. Each time they have a child there is a 50:50 chance that they will pass on the working copy and a fifty-fifty chance that they will pass on the faulty copy.

How do we know who has FAP?

Most people who have the gene for FAP begin to develop polyps in the colon in their early teens, so bowel checks should start between 10 and 14 years of age. Most people with FAP have developed polyps by the age of 30, and with every passing year without polyps, the likelihood that they have inherited the working copy of the gene increases.

The FAP gene has other effects on the body as well as causing polyps to appear. Some people get skin cysts, and some get harmless bumps on their bones.
For some reason, which we don't understand, most people get small black dots at the back of the eye which we call CHRPEs (this stands for congenital hypertrophy of the retinal pigment epithelium, but no-one ever calls them that!). These do not affect vision in any

way. A lot of people who do not have FAP have one or two of these black dots, but if there are more than five, or a large distinctive area, then this is a good indication that the person carries the FAP gene. If CHRPEs do not run in your family with polyps, then a clear eye examination does not mean that you do not carry the gene. There are different ways we can decide who has FAP; look for polyps, look for CHRPEs, do gene tests, or look for bony lumps or cysts.

The only way we can be sure that anyone has FAP is if we find a lot of polyps, or if we can prove that a person carries a faulty copy of the gene.

Gene Tests

If we think of a gene as a long word carrying a message, then the FAP gene has nearly 9000 letters in it. In different families it is a different letter or group of letters which have been wrongly copied which causes the gene not to work. The genetics team will try to work this out for each family, and if we find the "spelling" mistake we will offer a test to the rest of the family to find out who has it and who has not.
Blood cells contain copies of all your genes, so we can test the FAP gene from a blood sample. If a blood sample is not possible, saliva or other samples can sometimes be used.

Those who do not have the faulty copy of the gene can stop bowel screening, and they cannot pass FAP to their children. Those who do have the faulty copy will continue with regular screening as before, until polyps are found.

For some families we cannot offer a gene test. This may be because we have not yet found the fault in the gene in that family, or it may be that no one is available who has FAP to test. If no gene test can be done, you will be advised to carry on with regular bowel checks.

Looking for polyps

A short tube can be passed through the back passage into the lower part of the bowel. This examination is called a **sigmoidoscopy**, and you will be able to go home from hospital on the same day.

A longer, flexible tube can be used to examine more of the bowel, and this procedure is called a **colonoscopy.**

Doctors may use either method, but for colonoscopy, it is necessary to prepare the bowel by emptying it before the test. The hospital will give you details about this, but it usually involves taking a liquid medicine the day before the check, and sticking to a low bulk diet for a few days to ensure that the bowel is quite empty.

What happens if polyps are found?

Usually the doctor will take a sample of the lining of the bowel, or from a polyp, to be examined under the microscope. If these are harmless, it is sometimes reasonable to carry on with regular checks. However, most doctors offer the choice of having an operation if

they find a large number of polyps. Many people choose to have an operation as soon as polyps are found, rather than wait until there are large polyps.

The three main operations to remove the colon are:-

An **Ileo-rectal anastomosis (IRA)** involves removing the large bowel and attaching the small bowel to the rectum. This allows you to go to the toilet normally, but sometimes this can be many times a day. The rectum, which remains intact, may develop polyps so this must be examined at least once a year to prevent a cancer starting there.

A **pouch** operation involves the removal of the lining of the rectum when the colon is removed. A pouch is then made from the end of the small bowel, which then takes over some of the work of the colon. No checks have to be made on the rectum as there is no lining to grow polyps.

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.

Where else could problems occur?

People with FAP also develop polyps in the small intestine. Screening by endoscope from 25 will detect polyps and guide further management. Between 4 and 12% of patients develop cancer of the small intestine.

Is there a cure?

Research is ongoing to try to develop new treatments.

Doctors are also looking at non-surgical ways to stop polyps growing, such as drug treatment, and these may be recommended in certain circumstances.
These options may offer an opportunity to treat polyposis without surgery in the future but the only safe option at the moment is an operation.

Explanation of unfamiliar words:

FAMILIAL Something which runs in families.

POLYP A non-cancerous lump on the bowel wall.

ADENOMA (Adenomatous) There are different kinds of polyp: this describes how the polyps in FAP look under the microscope.

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

GENE One of the chemical recipes which control the working of the body.

DOMINANT Only one of a pair of genes is faulty.

CHRPE (Congenital hypertrophy of 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 seen.

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

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

ILEO-RECTAL ANASTAMOSIS (IRA) The small bowel is attached to the rectum when the colon has been removed.

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

PAN-PROCTO COLECTOMY (PPC) The colon and rectum are removed and the small bowel is brought to the surface of the abdomen.

ILEOSTOMY When someone has a PPC operation, the waste material is collected in a disposable bag.

DESMOID A non cancerous tumour which may grow in the abdomen leading to pain and obstruction.

Who should I contact for further advice?

The first person you should ask if you have any health problems is your family doctor. It may be that he/she is not very familiar with this problem, so you should take along this fact sheet. If anyone in your family has FAP, then they will have a surgeon, and you could ask them for advice. Every region in the UK has a genetics service and they can be contacted for help via your GP or directly for advice.

If you need more advice about any aspect of FAP, you are welcome to contact:

Cheshire and Merseyside Clinical Genetics Service
Liverpool Women's Hospital NHS Foundation Trust
Crown Street
Liverpool

L8 7SS

Telephone 0151 802 5003 or 5008

Facsimile: 0151 702 4286

This leaflet can be made available in different formats on request. If you would like to make any suggestions or comments about the content of this leaflet, then please contact the Patient Experience Team on 0151 702 4353 or by email at pals@lwh.nhs.uk

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