

Familial Adenomatous Polyposis (FAP)

A guide for people with FAP, their family and friends

This booklet aims to help you and your relatives understand Familial Adenomatous Polyposis (FAP), a rare condition that tends to run in some families.

The information in this booklet intends to add to, and not replace, discussions with doctors, genetic counsellors, nurses and other health professionals.

We suggest that you read this booklet in the order in which it is written, as each new section builds upon information in previous sections. Some medical terms that may be unfamiliar are explained in the glossary at the end.

What is FAP?

Familial adenomatous polyposis (FAP) is a rare inherited condition in which people develop many polyps (usually more than 100) mainly in their large bowel (colon and rectum). People with FAP are at very high risk of developing bowel cancer unless preventative actions are taken.

The name explains what it is:

Familial – that is, it runs in families.

Adenomatous – this is a term that describes the appearance of the polyps when examined under the microscope. This type of polyp has the potential to develop into cancer.

Polyposis means a large number of polyps. Polyps vary in size from a pinhead to a small mushroom (2 centimetres or more). Most people with FAP develop bowel polyps during their late teens. The polyps may start at any age, but rarely before the age of 10 years.

If not managed properly, FAP almost always leads to bowel cancer. This usually happens from the late 20s onwards.

You have a good chance of not developing bowel cancer if certain steps are taken. These involve removing most of the bowel before a cancer develops.

The bowel

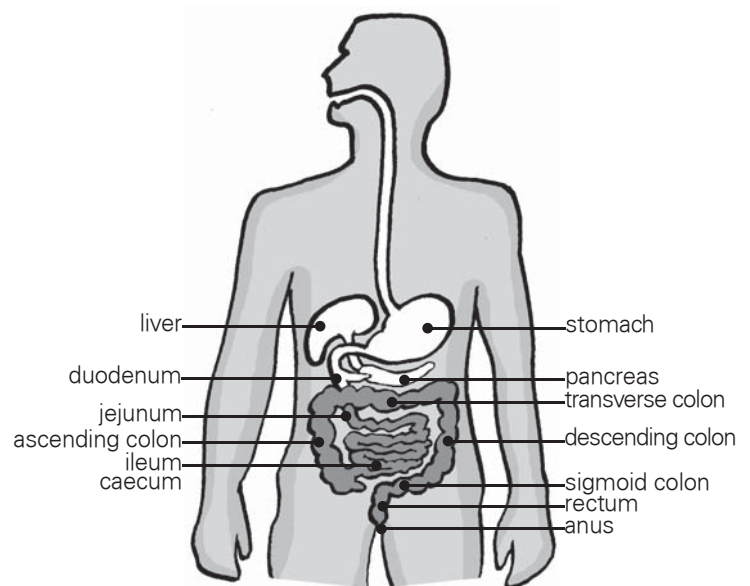
The bowel is part of the digestive system, or gut.

The gut is the long tube that runs from your mouth, via your stomach and bowel, to your back passage (anus). Food passes through the bowel, is digested and absorbed and the waste products are passed out as bowel motions.

The bowel includes:

- The small bowel or intestine (the duodenum, jejunum and ileum) where food is digested and absorbed.

- The large bowel (the colon and rectum) where only water and salts are absorbed. The colon has different sections – caecum, ascending colon, transverse colon, descending colon and sigmoid colon. The rectum leads to the outside of the body via the anus.



What causes FAP?

Every cell in the body carries a full set of instructions for growth and development called genes. Occasionally some genes do not work properly because there is a change in them. This is called a mutation.

FAP is caused by a mutation in the APC (Adenomatous Polyposis Coli) gene.

How is FAP inherited?

Genes come in pairs. You inherit one copy of every gene from your mother and the other copy from your father. This is how a gene mutation can be passed on in a family.

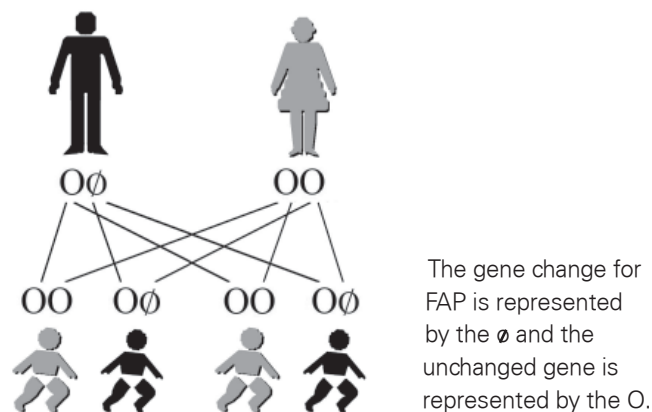
If someone carries a FAP mutation, each of their children, regardless of their sex, has a 50% (one in two) chance of inheriting the mutation. This does not mean that 50% of the children will necessarily be affected, but that each child has a 50% chance of being affected.

This diagram shows the chance that each child has of inheriting the mutation. When a baby is conceived, each parent passes on one copy of each of their genes to the baby. When one of the parents is carrying a mutation in one of the FAP genes, represented here by the “ø” symbol,

we see that there is a 50% chance that the baby will also carry this mutation.

If a person does not inherit the family's FAP mutation, then they cannot pass it on to their children.

If someone has FAP, it is possible that their brothers and sisters also have the condition. One of their parents could have the condition but may not know about it because they have not had any symptoms. This family should seek advice from genetic counselling professionals who know about FAP and can advise the family on screening and testing.



If someone has FAP but nobody else in the family has it, how did this happen?

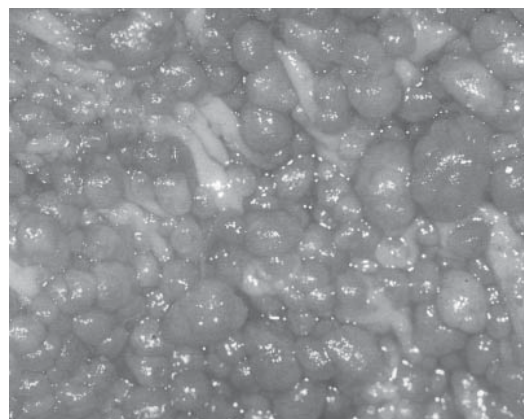
About one in every five people with FAP has no known family history of the condition.

A person may be the first person in the family to have FAP. Both parents may have passed on a working APC gene, but one of them, during development, may have then changed into a non-working APC gene. This change is called a new mutation.

It is possible that one parent had FAP but it was never diagnosed.

How do I know if I have FAP?

FAP can be diagnosed by an examination of the bowel and/or by genetic testing. When a doctor looks at your bowel, they will be looking for polyps such as in the picture below.



Polyps in the bowel, seen during a colonoscopy examination.

Other signs of FAP

CHRPE

CHRPE (pronounced 'chirpee') stands for Congenital Hypertrophy of the Retinal Pigmented Epithelium. Many people with FAP have pigmented patches (like freckles) on the innermost coat at the back of their eyeball (retina). These are called CHRPE. CHRPE can help doctors to discover whether FAP is present, because the pigmented patches can be detected at a young age, before polyps develop. To see them, an eye doctor needs to put drops in your eye, and then look into the eye with special equipment. Not everyone with FAP has CHRPE, and not everyone with CHRPE has FAP.

CHRPE is not a problem – it doesn't affect vision.

Polyps in other places

Polyps often develop in other parts of the gut such as the stomach or the duodenum (see the diagram on this page). Polyps in the duodenum may sometimes develop into cancer. The risk of cancer in the duodenum is much lower than that of colon cancer and usually occurs later than colon cancer.

A person with FAP needs to be checked regularly for these polyps by upper gastrointestinal endoscopy (gastroscopy). There are symptoms to watch out for: if you have indigestion, pain or weight loss you can't explain, if you are bleeding from the bowel or passing black bowel motions, or if the whites of your eyes go yellow (jaundice), tell your doctor about it.

If the polyps turn into cancer, the treatment is surgery. But research is still being carried out to decide whether, and how, duodenal polyps themselves should be treated if they are not cancerous.

Desmoid tumours

Some people develop desmoid tumours. These are fibrous lumps, usually found within the abdomen or in scars. They are often slow-growing, and may not produce symptoms for many years. Desmoid tumours can become a significant problem as they get larger, particularly if they constrict other structures such as blood vessels or bowel. They are not cancers and do not spread to other parts of the body.

What can be done if someone has FAP?

There is much that can be done if someone has FAP. This includes genetic testing, surgery, regular check-ups to look for polyps, and early treatment when polyps appear, before they turn into cancer.

Genetic testing for FAP

Genetic testing examines the information in a gene. Genetic testing can identify gene mutations.

Genetic testing involves taking a blood sample from an affected family member and sending it to a laboratory for

testing. Trying to find the FAP mutation is complex and time-consuming. The particular mutation in the APC gene varies from one family to another. The first step in each family is to try and find the family-specific FAP gene mutation by first testing a blood sample from an affected person.

This process of a “mutation search” may take considerable time, and it is not always possible to find the mutation that causes FAP for every family. In this situation the testing of other family members is not possible. Family members remain at risk of developing FAP and will need to undertake a programme of surveillance for the early detection of polyps and prevention of bowel cancer.

If, however, the family-specific mutation can be identified, then other at-risk relatives can have a test to see if they have inherited the FAP mutation. This is called predictive testing. Results are available more quickly because we already know the family’s specific FAP mutation. Genetic testing usually involves giving a small blood sample after written, informed consent is provided. Genetic counselling is offered before and after genetic testing by your Familial Cancer Clinic.

Regular check-ups

People with a clinical diagnosis of FAP, an abnormal gene test or family members at-risk of FAP may have:

- Colonoscopy every year to check for polyps from age 10–16 years (dependent on the history of FAP in the family). When polyps appear, discuss surgery with your doctor.
- Removal of the bowel (colectomy, see page 13) once polyps are found. The aim is to do this as preventative surgery, before a cancer develops.
- Regular (six monthly or yearly) examination of any remaining bowel or rectum following colectomy.
- Regular endoscopy of the stomach and duodenum.

Tests used during regular check-ups

Your doctor will advise which is the most suitable test for you.

Colonoscopy

A colonoscopy examines the inner lining of the large bowel. A flexible tube with a light at the end is passed through the anus and moved through the bowel to enable the doctor to see along its full length. The bowel has to be empty, so some medication is usually given the day before surgery to help you to empty your bowel. The examination generally takes up to 30 minutes. It is done under sedation as a day procedure. If any polyps are seen, they can usually be removed or sampled by biopsy at this time.

Sigmoidoscopy

A sigmoidoscopy involves the gentle insertion of a narrow lighted tube through the anus to view the inner lining of the lower part of the large bowel. It does not require any

sedation and takes only a few minutes. It only examines the very end of the bowel, and can be used to examine the residual bowel after bowel surgery.

Upper gastrointestinal endoscopy

An upper gastrointestinal endoscopy examines the inner lining of the upper part of the gut (e.g. stomach and duodenum) using a similar type of technology to the colonoscopy. The tube in this case is inserted through the mouth. Light sedation is often given to the patient. To ensure the stomach and upper gut are empty, no food or drink should be taken for several hours before the procedure.

Biopsy

Sometimes a small sample (biopsy) of the lining of the bowel is taken for examination under the microscope to check if there are microscopic changes. This is done through the colonoscope and is not painful.

Affected and at-risk people should immediately tell their doctor or Familial Cancer Clinic about any symptoms of bowel problems such as rectal bleeding, indigestion, abdominal pain, changes to bowel habits (like prolonged constipation or diarrhoea) or genito-urinary symptoms such as blood in the urine or abnormal bleeding from the vagina. In general, any persistent new symptoms should be discussed with your doctor or Familial Cancer Clinic.

Surgery

The treatment for FAP is a preventative operation to remove all or most of the large bowel (the colon and sometimes the rectum) after bowel polyps appear, but before cancer occurs.

The aim is to reduce the risk of cancer developing and to keep your digestive system working as normally as possible. Losing your large bowel does not affect how your body absorbs food.

It is known that people who put off surgery are much more likely to develop cancer and die at a young age than those who have surgery before the onset of bowel cancer.

Talk to your surgeon about what is involved. You may find it helpful to talk with someone who has had the same operation. Your surgeon, counsellor or the Hereditary Cancer Registers may be able to arrange this.

Operations for FAP

Operations available for FAP include:

- colectomy and ileo-rectal anastomosis
- restorative proctocolectomy
- proctocolectomy and ileostomy.

Each has advantages and disadvantages. Deciding on the best option is not always straightforward.

You should talk to your surgeon about the best treatment for you.

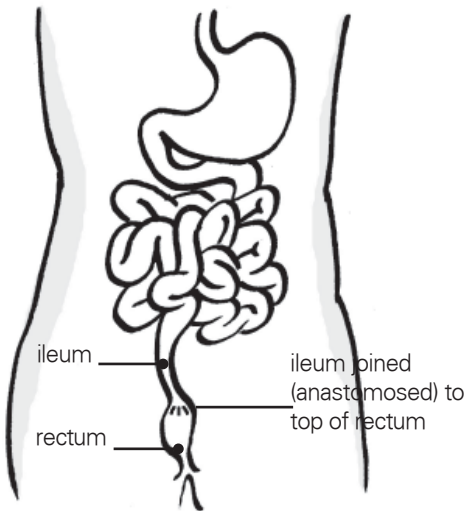
Colectomy and ileo-rectal anastomosis

All of the colon is removed (colectomy), and the end of the ileum is joined to the rectum (ileo-rectal anastomosis or IRA).

After recovering from surgery, most people find they have two to three bowel actions a day.

The advantage is that you keep the rectum and anus, so you can continue to pass bowel motions in the usual way.

The disadvantage is that there is still a risk of polyps and cancer in the rectum. You will need regular check-ups for the rest of your life to remove any small polyps in the rectum before they become a problem.



A total colectomy involves removal of all the colon

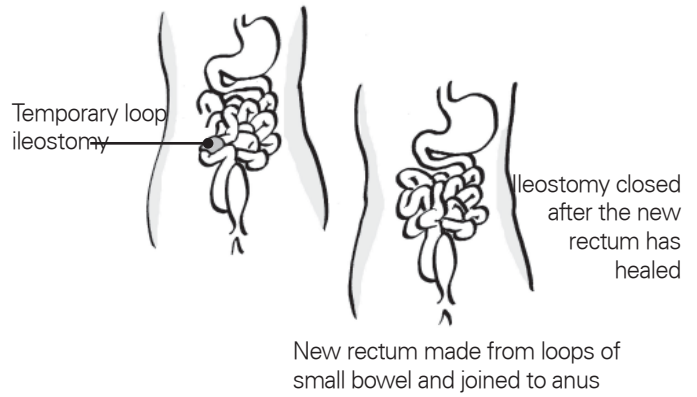
Restorative proctocolectomy (the pouch procedure)

All of the colon and the rectum and all or most of its lining is removed.

A new “rectum” is made from the end of the small bowel. This is called a pouch and is joined to the anus. You may have a temporary opening through the abdominal wall (ileostomy) so that the bowel contents can leave your body by an alternative route for about three months while the pouch surgery heals.

The advantage is that the risk of cancer developing in the rectum is less than for the IRA operation. With the lining of the rectum gone, there is less danger of polyps or cancer forming there. After the temporary ileostomy has gone, you will go to the toilet to pass bowel motions in the usual way.

The disadvantage is that some people have more frequent bowel motions during the day. Occasionally there may be some leakage, especially at night. Other problems, such as post-operative inflammation, infection and adhesions (loops of remaining bowel sticking together) are also more common. Although the risk of cancer in the pouch is low, it can occur.

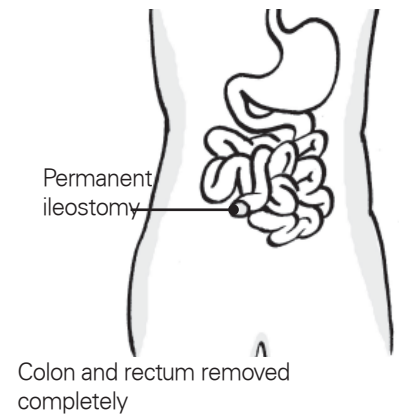


Proctocolectomy and ileostomy

This is rarely done now. It may be done if there is already a cancer low in the rectum or if there are significant problems after one of the other two operations.

All of the colon, rectum and anus are removed in this operation. The end of the small bowel (ileum) is then joined to the wall of the abdomen, opening to the outside to allow bowel motions (faeces) to pass out. The opening is called the stoma.

A special bag is worn over the stoma to collect the bowel motions.



What can I expect after treatment?

After the operation

The main difference for most people is simply more frequent bowel motions. If you do have problems, for instance in managing diarrhoea, there are many approaches that can help. Talk these problems over with your doctor.

After a few months, your bowel usually gets used to being shorter and adapts. Various medicines can help control the diarrhoea and can be useful if you are under stress or need to go to a special event.

Regular check-ups

After the operation, you will still need regular check-ups with your doctor.

Relationships, sex and bowel surgery

On the physical side, surgery for FAP, even having an ileostomy, should not affect your ability to enjoy sex. For some people, however, surgery can change the way they feel about themselves and their bodies. Or they may be concerned about how their partner will cope and the

longer term impact of surgery on their relationship with their partner.

It can help considerably to discuss these issues with someone else. Another person can see your situation differently and may suggest new ways that you might manage it. This person may be your doctor, a stomal therapist, another family member or health professional. There are also experienced counsellors who specialise in issues around sexuality who could be of assistance. **The Cancer Council Helpline 13 11 20** can provide you with further information.

Pregnancy and childbirth

Pregnancy and childbirth may pose problems for some women following bowel surgery. Women who have had a restorative proctocolectomy may find it harder to fall pregnant. This can be important if a woman needs to have surgery before she has completed her family. Pregnancy rates may be less affected by other types of bowel surgery.

Some time should be allowed between having surgery and becoming pregnant and there may be some types of bowel surgery where a caesarean delivery would be advised. These matters should be discussed with your doctor or obstetrician, as each situation will be different.

Support and information

For further information and support about FAP in your family please contact:

The Familial Cancer Unit
Child, Youth and Women's Health Service
Women's & Children's Hospital
72 King William Street
North Adelaide SA 5006
t 08 8161 6995
f 08 8161 7984
cywhs.famcancer@cywhs.sa.gov.au

Glossary

adenoma

A non-cancerous tumour. It may turn into a cancer if not treated.

anastomosis

The joining together of two tubes, such as two cut ends of the bowel.

anus

The back passage. The opening of the bowel through which bowel motions are passed.

benign

Not cancer.

CHRPE

Harmless pigmented patches inside the eyeball on the retina. They are common in FAP. CHRPE stands for congenital hypertrophy of the retinal pigmented epithelium.

colectomy

The surgical removal of the colon.

colon

The part of the large bowel between the end of the small intestine (the ileum) and the rectum. It is about 1.5 metres long.

colonoscopy

Examination of the large bowel using a thin flexible tube with a light at the end, called a colonoscope. It is passed through the anus and gently moved around so that, the doctor can see the full length of the large bowel.

desmoid tumour

A mass of fibrous tissue.

duodenum

The first 30 centimetres of the small bowel. The stomach empties into the duodenum.

faeces

Bowel motions; waste matter passed from the gut via the back passage (anus).

gene

The elements of a cell that carry instructions on how the cell should grow and function. Each person has a set of many thousands of genes inherited from both parents. This set is found in every cell of the body.

ileostomy

An opening through the abdominal wall through which the ileum is brought to replace the function of the anus. An ileostomy may be performed after the surgical removal of the colon and rectum. It may be temporary or permanent.

ileum

The lower half of the small bowel, which joins up with the colon.

jaundice

A yellowish staining of the skin and the whites of the eyes.

jejunum

The part of the small bowel below the duodenum and leading into the ileum.

large bowel

The colon and rectum.

malignant

Cancerous. Malignant cells can spread (metastasise) if not treated.

mutation

A change in a gene causing it to show a new characteristic.

polyp

An abnormal growth or lump in the bowel, often on a stalk, like a mushroom. Polyps are usually benign but can turn cancerous.

polyposis

The condition of having large numbers of polyps in the large bowel.

rectum

The last 12-15 centimetres of the large bowel, which opens to the outside of the anus. The faeces collect in the rectum before they are passed as a bowel motion.

retina

The light-sensitive lining inside the eyeball.

sigmoid colon

The last 20-25 centimetres of the colon, which leads into the rectum.

sigmoidoscopy

Examination of the rectum and sigmoid colon using a sigmoidoscope. This is a narrow lighted tube. It is inserted gently through the anus, and gives a view of the lining of the bowel.

stoma

An artificial opening created in the body by surgery.

From The Cancer Council South Australia

The Cancer Council fosters all aspects of the fight against cancer and is involved in the support of all who may be affected directly or indirectly by cancer. Contact **The Cancer Council Helpline 13 11 20**:

The Cancer Council South Australia
202 Greenhill Road, Eastwood
PO Box 929, Unley, SA 5061
t 08 8291 4111
f 08 8291 4122
chl@cancersa.org.au
www.cancersa.org.au

This booklet is one of a series of resources produced by The Cancer Council to help you understand more about your illness and to help yourself. Other titles available are:

About cancer
About chemotherapy
About surgery
Cancer information on the internet
Caring for the person with advanced cancer
Clinical trials
Clinical trials an overview
Emotions and cancer
Guide for partners of women with breast cancer
Guide to cancer services in Adelaide
Hair loss
How can I relax?
I want to help
Making choices - Alternative and complementary therapies
Nutrition for people having cancer treatment
Oral health during cancer treatment
Questions you might like to ask your doctor
Sexuality for men with cancer
Sexuality for women with cancer
Skin care during cancer treatment
Understanding and controlling cancer pain
Understanding radiation therapy
What About Me? (for children when a parent has cancer)
What do I eat now?
When you're diagnosed with cancer

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