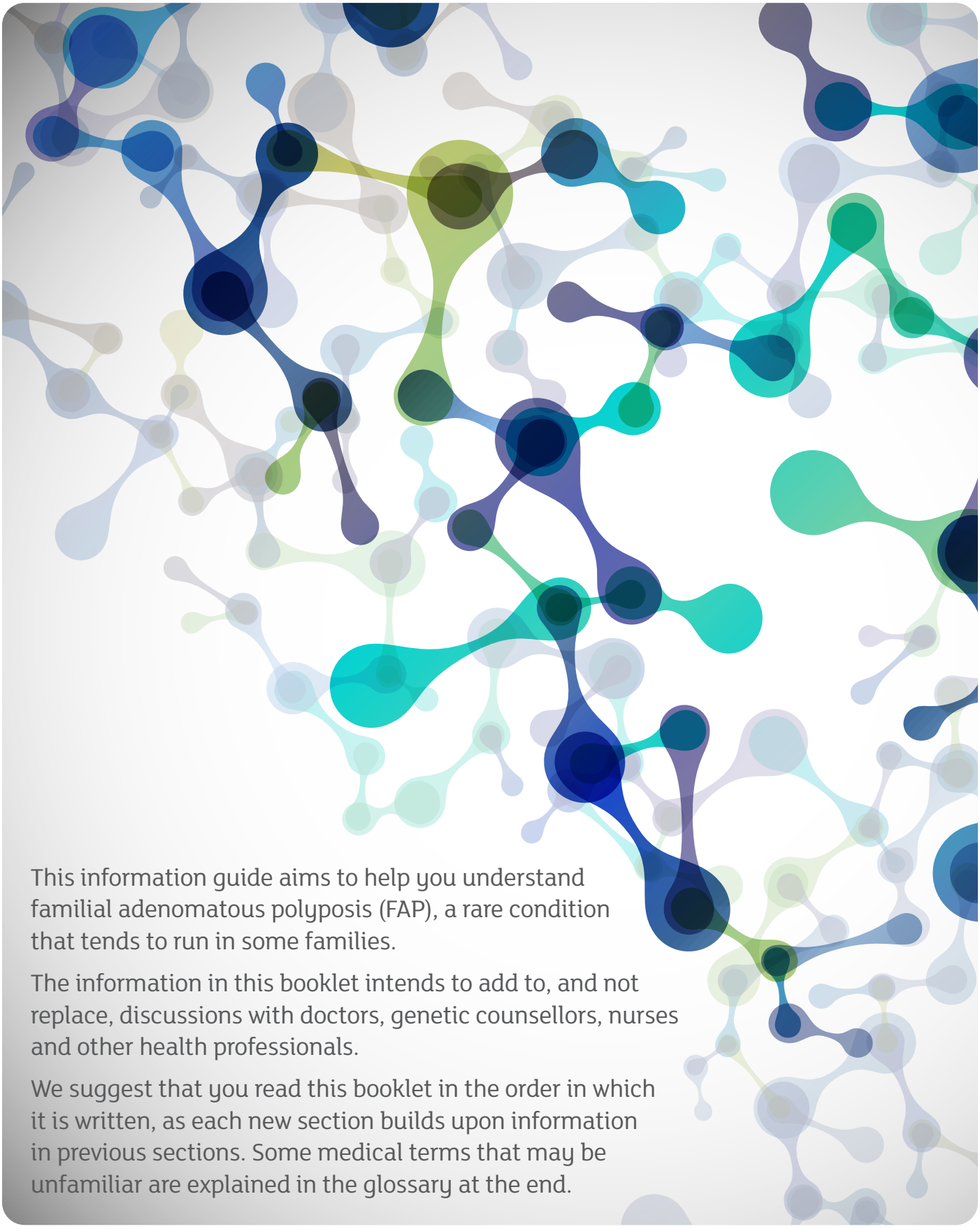
Familial adenomatous polyposis (FAP)

A guide for people with FAP, and their family and friends



This information guide aims to help you 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.

1. 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.

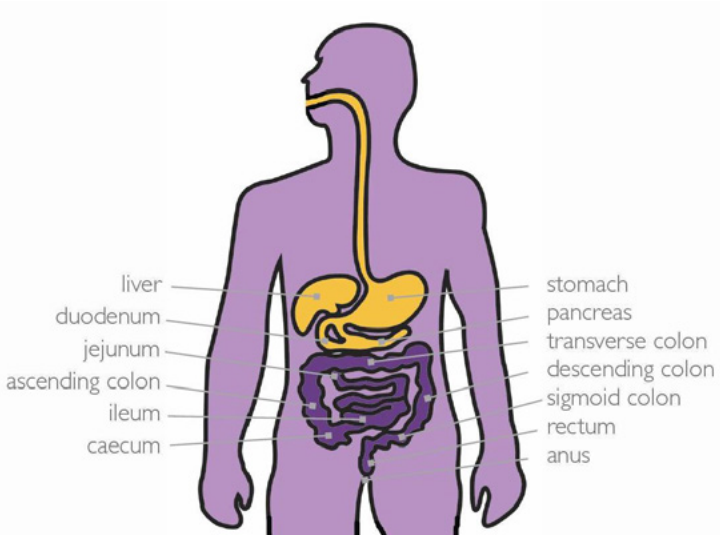
If not managed properly, FAP almost always leads to bowel cancer. This usually happens from the late 20s onwards.

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.

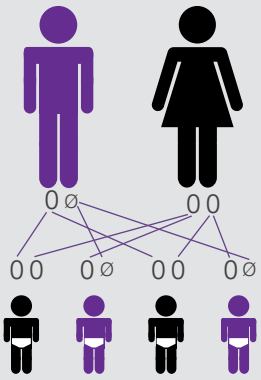
How is FAP inherited?

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 FAP mutation, then they cannot pass it on to their children.



The gene change for FAP is represented by ø and the unchanged gene is represented by the 0.

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?

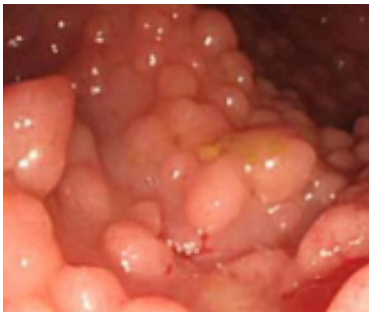
About one in every five people with FAP has no known family history of the condition.

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 these gene copies, during the baby's development, may have then changed into a faulty APC gene. This change is called a new mutation, or 'de novo' mutation.

It is also 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 to the right.



Polyps in the bowel of someone with FAP, 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 develop 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 often appear 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 page 2). 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 at a later age 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 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. They are not cancers and do not spread to other parts of the body. Desmoid tumours can become a significant problem as they get larger, particularly if they press on other structures such as blood vessels or the bowel.

2. 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 involves taking a blood sample from an affected family member and sending it to a laboratory for testing. Trying to find the APC mutation which causes FAP is complex and can be time-consuming.

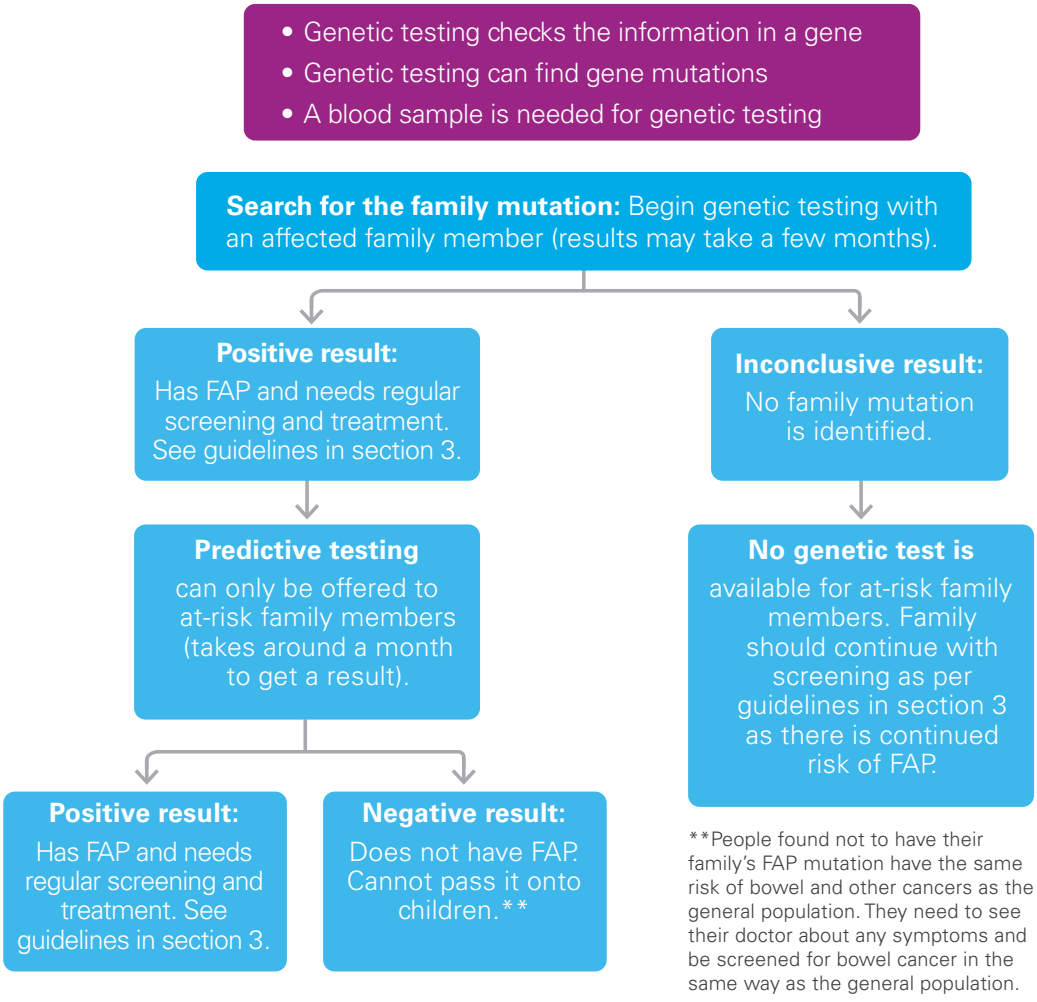
The particular mutation in the APC gene varies from one family to another. The first step in each family is to try and find the family-specific mutation by first testing a blood sample from an affected person. This process of a "mutation search" may take considerable time, and it is not always possible to find the mutation that causes FAP for a family.

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 APC mutation. This is called predictive testing. Results are available more quickly because we already know the family's specific APC mutation. Genetic testing usually involves giving a small blood sample after written consent is provided. Genetic counselling is offered before and after genetic testing by your Family Cancer Clinic.

If you have inherited the APC mutation, you have or will develop FAP. If you have not inherited the mutation, then you do not have FAP.

If you have not inherited the family mutation, then you do not need surgery or intensive screening and your children are not at risk of inheriting FAP from you.

Genetic testing in Australia is done through Family Cancer Clinics, where professionals make sure that people receive all the support and help they may need to make decisions about testing. Genetic testing done through a Family Cancer Clinic is free.



**People found not to have their family's FAP mutation have the same risk of bowel and other cancers as the general population. They need to see their doctor about any symptoms and be screened for bowel cancer in the same way as the general population.

Genetic testing: Frequently asked questions

Can all families with FAP have the gene test?

Not all families can have the FAP genetic test because:

- FAP genetic testing is only offered to families when the family history suggests the possibility of FAP.
- Sometimes a family thought to have FAP has no living affected family member to test or has some affected individuals who are not interested in having genetic testing. This means that the family-specific mutation cannot be identified, so predictive testing is not available to other at-risk members of the family.
- In some families, even though a blood sample is available, the laboratory may not be able to find the family-specific APC mutation. This may be due to a type of mutation that is difficult to find and/or the possibility of a genetic change on an as yet unidentified gene associated with FAP.

What happens if I can't be gene tested?

If you are at risk of FAP, you will need to have regular bowel examinations to check for polyps. If many polyps are found the diagnosis of FAP may be made on these results (this is called a clinical diagnosis). Your treatment options then are the same as for everyone else with FAP.

What if I don't have the family's FAP gene mutation?

If you don't have the gene mutation found in other members of your family, you do not have FAP and do not have a high risk of developing bowel cancer. Your children are not at risk of inheriting FAP because you don't have the mutation to pass it on to them. They do not need to have the gene test. You and your children no longer need to have screening for FAP. A negative FAP gene test does not mean that you cannot develop bowel cancer but that your chance of developing bowel cancer is the same as for the general population – about a 1 in 20 chance in your lifetime.

What if I do have the family FAP gene mutation?

If the test finds that you carry the APC mutation, then you have or will develop FAP. It is vital that you have regular screening of the bowel and rectum and have treatment when polyps appear, before bowel cancer develops. For details about regular screening see page 11.

When can the test be done?

Once the FAP mutation in your family has been identified, it is possible for other at-risk family members to also have genetic testing. Genetic clinics usually prefer young people to have a say in whether they want the test or not. This means leaving their test until the teenage years. If you are concerned about this, talk it over with your doctor or genetic counsellor at the clinic. However, if symptoms like rectal bleeding or abdominal pain occur, then testing might be done earlier.

Who will know my test result?

Your personal test result is shared only with persons or organisations for whom you give consent.

The importance of talking it over

Before you have a genetic test, it is important to talk with a professional such as your doctor or genetic counsellor, about the pros and cons of genetic testing. This may help you decide if genetic testing is right for you.

Insurance policies and genetic testing

If you are planning to have genetic testing, it is wise to discuss insurance issues with a genetic counsellor before testing.

I'm not sure I want to know.

This is a common reaction. It may be helpful to consider the following points:

- If the test shows you do not have the family FAP mutation, you will know that you will not develop cancer due to FAP. This means you have the same risk of developing bowel cancer as the general population. You will not need to have screening until the age recommended for the general population, unless symptoms arise.
- If the test shows you have the family mutation, regular screening and surgery to remove the bowel greatly reduces the risk of developing cancer.

3. Regular check-ups

People proven to have FAP

People with a clinical diagnosis of FAP or a positive genetic test may have:

- Colonoscopy every year to check for polyps. When polyps appear, discuss surgery with your doctor.
- Removal of the bowel (colectomy, see page 7) should usually be performed by age 20. 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.
- Regular endoscopy of the stomach and duodenum.

Relatives of those proven to have FAP

If the family mutation **has** been identified, genetic testing (see page 4) may be offered to all at-risk relatives. Then:

- if the person is found not to carry the family mutation, screening is not required
- if the person does carry the family mutation, screening and treatment is recommended.

If the family mutation **has not** been identified (predictive testing not possible), then:

- start screening from age 10-15 years with colorectal examination every year until the age of 35
- if no polyps have developed by age 35 years, reduce colorectal examination to every three years
- at 55 years of age, if polyps have still not developed, screening for colorectal cancer can revert to that of the general population.

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 the colonoscopy 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 sigmoidoscope and should not be painful.

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

4. 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, genetic counsellor or the Hereditary Cancer Registry may be able to arrange this.

Operations for FAP

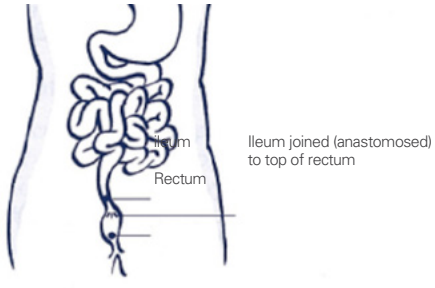
Operations available for FAP include:

- colectomy and ileorectal 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 ileorectal 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

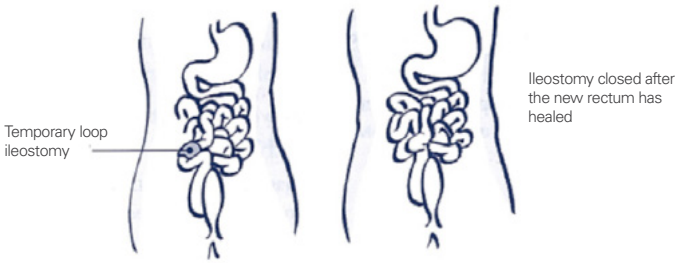
Restorative proctocolectomy (the pouch procedure)

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.



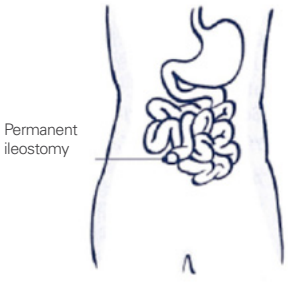
New rectum made from loops of small bowel and joined to anus

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.



Colon and rectum removed completely

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.

A family's story

Annette was 14 when her mother, Joan, was found to have FAP and bowel cancer. Joan, 36, was treated for the cancer but lived only another two years.

Checking back, Joan and her doctor realised that both Joan's father and grandmother had died of bowel cancer at a young age. It was clear that this was a family disorder. So Annette and her brothers were examined. She and a younger brother, Rob, were found to have polyps.

At the age of 16, Annette had a total colectomy and ileorectal anastomosis. After joining the Hereditary Cancer Registry she received annual reminders for her check-ups. She has had yearly check-ups with sigmoidoscopy ever since, and remains fit and healthy. Rob had the operation three years later, and he too remains well.

Joan and her parents and grandparents did not know about FAP and their risk of inheriting it, and so were never checked for it. It was only Joan's bowel cancer that brought it to light.

The younger generations are luckier. When Joan was diagnosed, her sister Peg and Peg's children were also checked. Peg and her son Paul (Annette's cousin) were found to have FAP. Both have had bowel surgery. So far, however, Peg's other two children (both now in their thirties) have not developed polyps – they probably don't have FAP.

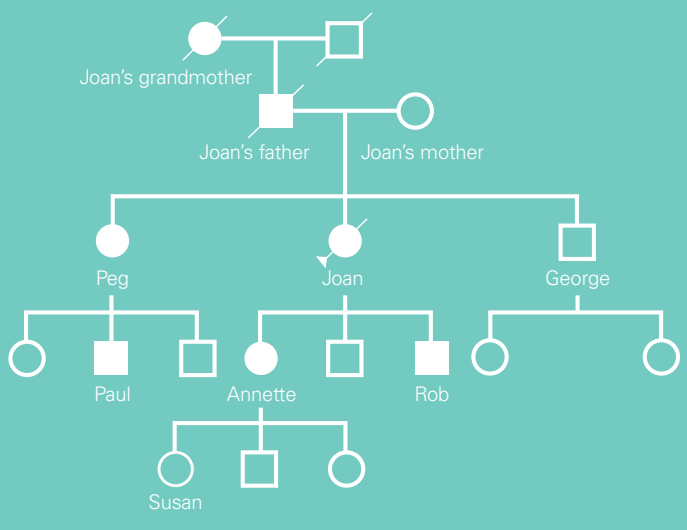
Joan's younger brother, George, did not want to know about it and never went for check-ups with the doctor. So far he has been lucky – he is now nearly 50 and remains healthy. He probably doesn't have FAP – but it is still possible that he does and that polyps will develop into bowel cancer. His two teenage daughters still don't know whether they are at risk.

Annette is now married with three children. Only recently, Annette saw a Family Cancer Clinic who recorded her family history and arranged for genetic testing. Annette had a blood sample taken to identify the family FAP gene mutation. She has discussed FAP with her children. Her eldest daughter, Susan, now 12, has decided to have the gene test between her 13th and 14th birthdays.

Once the gene mutation that causes FAP has been identified for this family, genetic testing can also be offered to Peg's other children, and even to Peg's brother George. If George does not have the mutated gene then he and his daughters will never develop polyposis.

Annette's story highlights the importance of knowing your family health tree. Drawing your extended family tree, and obtaining information about the family history of bowel problems and cancer, can help to determine who is at risk of FAP in your family.

Annette's Family tree



5. Support and information

The NSW & ACT Hereditary Cancer Registry

The Hereditary Cancer Registry (HCR) was established to provide information and support to people affected by hereditary cancer, their family members who may be at risk, and their doctors in NSW and the ACT. While there are a number of hereditary related cancers, the conditions focussed on by the HCR include:

- Lynch syndrome (Hereditary Non-Polyposis Colorectal Cancer)
- Familial Adenomatous Polyposis – FAP
- Peutz-Jeghers Syndrome – PJS
- Juvenile Polyposis
- Other polyposis syndromes (i.e. Serrated Polyposis, Mixed Polyposis, MYH Associated Polyposis).

What does the Hereditary Cancer Registry do?

The HCR aims to assist people from families with a high risk of hereditary cancer to understand and manage their risk. It does this by:

- Building a complete picture of the condition in a family, assisting doctors to assess risks and plan screening and treatment.
- Developing knowledge about the incidence of hereditary cancer conditions in NSW and ACT.
- Contributing to better information, services and support for patients and their families.
- Providing a screening reminder service to registrants and their doctors.

The HCR offers an optional screening reminder service to assist registrants with keeping up to date with screening appointments. The HCR records registrants' contact details, family history, treatment, screening test results and details of any surgery that has been performed. All the information kept at the HCR remains strictly confidential. It is not given to anyone else, including other family members, without permission.

The information on the HCR database is an excellent starting point for national and international research into hereditary cancer syndromes such as Lynch syndrome. Information that is recorded on people or families is only used in statistical reports in which individuals cannot be identified. The HCR receives advice from a committee of clinical experts.

Why should I register and how do I register?

People from high-risk families are encouraged to register with the HCR to take advantage of the up-to-date information, screening reminder service and support provided by the registry. Registration is voluntary, and personal information is held confidentially under the NSW Health Information and Records Privacy Act. Registration can be completed using the registration form provided by your doctor or genetic counsellor, or contact the HCR for more information.

Other services offered by the NSW & ACT Hereditary Cancer Registry include:

- A screening reminder service for you and your doctor/s to help you keep track of your various appointments.
- Information about hereditary cancer, its prevention, treatment and management.
- Booklets and pamphlets about hereditary cancer conditions.
- Information about genetic services and tests.
- Linking separate branches of an extended family.
- Assistance with informing relatives of their risk by providing you with letters and other information about the family condition to pass on to them.
- Assistance with contacting support groups (cancer/genetics/ostomy).
- Assistance with passing on your hereditary information (with your consent) to interstate and overseas hereditary registers when you move.
- Referral to confidential telephone counselling.

For more information, contact the NSW & ACT Hereditary Cancer Registry:

Phone: 1800 505 644 **Fax**: 02 8374 3644 **Email**: CINSW-HCR@health.nsw.gov.au

Mail: Cancer Institute NSW, NSW & ACT Hereditary Cancer Registry
Locked Bag 2011, St Leonards NSW 1590.

Web: www.cancer.nsw.gov.au/data-and-statistics/cancer-registries/hereditary-cancer-registry

5. Support and information

Support and information: Family Cancer Clinics		
The staff at Family Cancer Clinics are trained to assist you with concerns about your family's history of cancer. Family Cancer Clinic staff can undertake a family risk assessment, provide you with screening recommendations, organise for you and your family to be seen by specialists and, if appropriate, organise genetic testing.		
Camperdown	Hereditary Cancer Clinic Department of Cancer Genetics Level 2, Gloucester House Royal Prince Alfred Hospital Missenden Rd, Camperdown NSW 2050	(02) 9515 8780
Darlinghurst	Family Cancer Clinic Department of Medical Oncology Level 5, The Kinghorn Cancer Centre 370 Victoria Street, Darlinghurst NSW 2010	(02) 9355 5647
Kogarah	Genetic Counselling Service Cancer Care Centre, St George Hospital 1 Short Street, Kogarah NSW 2217	(02) 9113 3815
Newcastle	Hunter Family Cancer Service Cnr Turton & Tinonee Roads, Waratah NSW 2298	(02) 4985 3132
Randwick	Hereditary Cancer Clinic Prince of Wales Hospital High St, Randwick NSW 2031	(02) 9382 5107
St Leonards	Familial Cancer Service Dept Clinical Genetics Clinical Admin 3E Royal North Shore Hospital St Leonards NSW 2065	(02) 9463 1554
Westmead	Familial Cancer Service Crown Princess Mary Cancer Centre Westmead Hospital PO Box 533, Westmead NSW 2145	(02) 8890 6947

For locations of Family Cancer Clinics in Regional NSW:

Phone: 1800 505 644

Web: www.cancer.nsw.gov.au/learn-about-cancer/cancer-in-nsw/hereditary-cancers/cancer-genetics-counselling-services

An up-to-date list of Family Cancer Clinics can also be found on the Centre for Genetics Education website:

Web: www.genetics.edu.au/genetic-services/cancer-genetics-clinics

Other sources of support & information		
Centre for Genetics Education	Information fact sheets and other information about hereditary cancer is available from the Centre for Genetics Education.	(02) 9462 9599 www.genetics.edu.au
Cancer Council Helpline	Cancer Council Helpline is a telephone information service provided by Cancer Council NSW for people affected by cancer. For the cost of a local call, you can talk about your concerns and needs confidentially with oncology health professionals. Helpline consultants can also put you in touch with appropriate services in your area.	13 11 20 www.cancer.org.au/about-cancer/patient-support
Genetic Alliance Australia (formerly AGSA)	For information about support groups contact Genetic Alliance Australia.	(02) 9295 8359 www.geneticalliance.org.au
eviQ Cancer Treatments Online	eviQ Cancer Treatments Online is a website that provides health professionals with current evidence-based, peer-reviewed, best-practice cancer treatment and information. The Cancer Genetics section of eviQ provides information about hereditary cancer conditions such as FAP, including information sheets for people and families.	www.eviq.org.au (click on the Cancer Genetics tab)
Stoma Associations	If you have an ileostomy, your stomal therapy nurse can give you practical tips about how to cope with your ileostomy and which bags are best for you. An association such as Ostomy NSW Ltd or Colostomy Association of NSW Inc may also be helpful. Someone from the association who already has an ileostomy may visit you at home or in hospital. They can talk with you about your fears and worries and explain what works best for them. Many people find this very helpful. There are 22 regional, voluntary stoma associations across Australia which distribute stoma related products to their members.	Ostomy NSW Ltd (02) 9542 1300 www.ostomynsw.org.au Colostomy Association of NSW Inc 1300 678 669 www.nswstoma.com.au

6. 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 sigmoidsoscope. This is a narrow lighted tube. It is inserted gently through the anus, and gives a view of the lining of the bowel.

Small bowel
The first part of the gut made up of the duodenum, jejunum and ileum.

Stoma
An artificial opening created in the body by surgery.

This guide is based on the booklet *Understanding Familial Adenomatous Polyposis (FAP) Syndrome*, published by the Cancer Council NSW in June 2004.

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