

St Mark's Hospital at Northwick Park specialises in treating familial adenomatous polyposis (FAP).

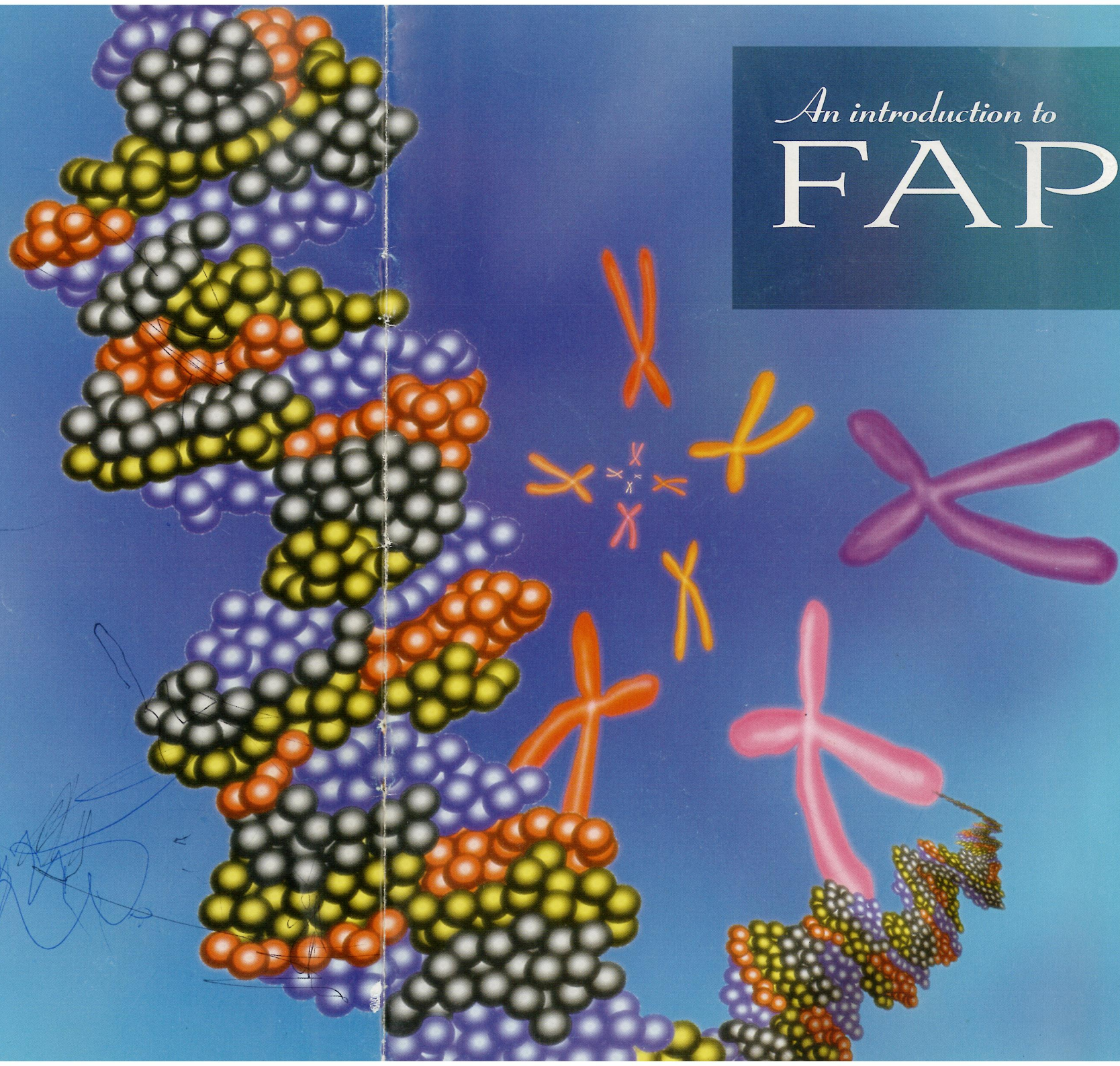
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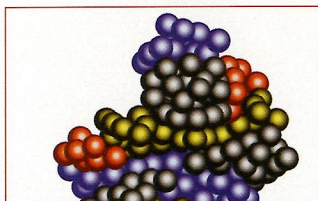
If you would like to make a contribution please make a cheque payable to The St Mark's Research Foundation (Polyposis Registry) and send it to The Polyposis Registry, St Mark's Hospital, Watford Road, Harrow, Middlesex. HA1 3UJ.

All donations are gratefully received and carefully used.

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An introduction to FAP





Your contacts will be:-

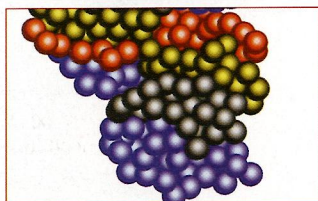
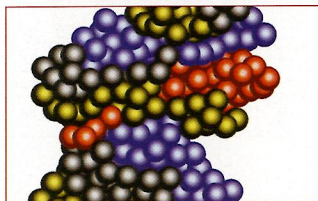
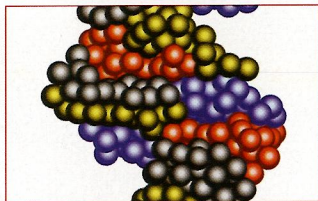
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nucleus the inner part of the cell which contains chromosomes

oesophagus the gullet

polyp a small swelling, arising from the lining of the bowel

rectum the last part (about 15cm) of the large bowel. It lies between the colon and anus

restorative proctocolectomy(pouch) the operation in which the colon and rectum are removed leaving the anus. An ileal reservoir or pouch is then made and joined to the anus

sigmoidoscopy an examination of the rectum and distal colon

small intestine the part of the intestine between the stomach and the colon which is responsible for digesting the food. It is divided into three parts: the duodenum, the jejunum and the ileum

stomach stores the food before it passes into the small bowel

symptoms what is complained about

The Red Lion Group

In 1994 this group was established to help people with, or about to have, a pouch by providing information and introducing individuals who have already had the operation. The group can be contacted through the pouch support sister at St Mark's Hospital. The telephone number is 0181 235 4110.

Glossary

<i>adenoma</i>	a polyp growing from glandular tissue in the mucous membrane of the bowel
<i>anal sphincter</i>	circular muscle at the anus which relaxes to allow faecal matter to pass through
<i>anastomosis</i>	a surgical join between two pieces of bowel
<i>anus</i>	the opening at the end of the gut, through which faeces are discharged
<i>autosomal dominant</i>	50:50 chance of inheritance
<i>biopsy</i>	a piece of tissue taken for examination under the microscope
<i>chromosome</i>	a length of coiled DNA.
<i>colectomy with ileo rectal anastomosis</i>	the operation in which the colon is removed and a surgical join made between the last part of the small bowel and rectum
<i>deoxyribonucleic acid (DNA)</i>	a complex chemical that contains the genetic code. Passed on from one generation to the next.
<i>duodenum</i>	first portion of the small intestine
<i>faeces</i>	waste matter discharged from bowels
<i>familial adenomatous polyposis</i>	an inherited condition in which more than 100 adenomas occur in the large intestine
<i>gastrointestinal tract</i>	the guts from mouth to anus
<i>gene</i>	a genetic building block
<i>ileostomy</i>	the ileum when brought out on to the abdomen
<i>ileum</i>	lower part of the small intestine
<i>large intestine</i>	the large bowel, the colon and the rectum



Familial Adenomatous Polyposis

This booklet sets out to answer some of the questions most often asked by people who have FAP and by their relatives.

If you have any questions or would like further explanations please do not hesitate to ask.

What is Familial Adenomatous Polyposis (FAP)?

FAP is an inherited condition which mainly affects the large intestine (also known as the large bowel or colon and rectum). A diagram of the intestine together with an explanation of how it functions can be found on page 6.

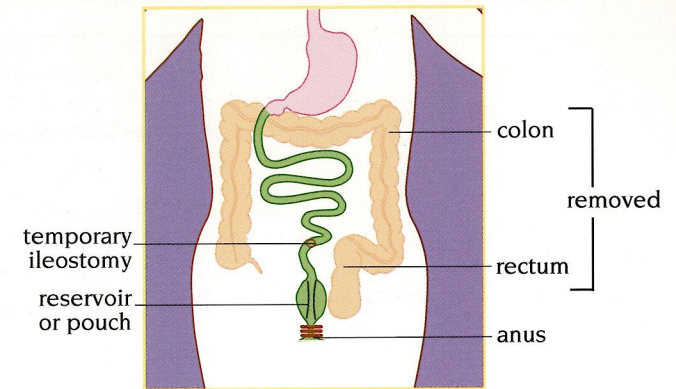
People with FAP develop many polyps (which are like small cherries on stalks) inside their large bowel. There are many different types of polyps but these particular polyps are called adenomas (the "adenomatous" in FAP). An adenoma can in time turn into a cancer which is why it is so important to make sure anyone at risk of inheriting FAP is examined. FAP is a serious condition unless detected early when it can be treated.



The faulty gene that causes FAP is carried on chromosome number 5.

What causes FAP?

FAP is a genetic condition, that is to say it is caused by a single faulty gene. The FAP gene is one of many thousands of genes carried on our chromosomes which determine our physical characteristics (our hair and eye colour, the shape of our nose, our blood type and the number of fingers and toes we have.) Genes carry the information required to determine these characteristics as a chemical code in the form of DNA (deoxyribonucleic acid).



Life after the operation

In the early stages most people pass soft stool. When things have settled down most people find they need to go to the lavatory between 4 and 6 times a day. The stool will be the consistency of porridge. It should be no problem to hold on for a while after feeling the urge to go. Some people need to go at night and a few may feel more confident if they wear a small pad in case of any minor leakage. Some people find that certain types of food upset their bowels but they find out which by trial and error. Foods that are fibrous or pithy (for example, oranges) should be eaten with care or avoided as they may lead to the bowel becoming obstructed. If a person's bowel actions are too frequent they may need to adjust their diet or take tablets such as loperamide (Imodium). These tablets slow down the bowel and are not addictive.

Follow up after surgery

When everything has settled down after the operation, it is usual to return to the outpatient clinic once a year.

This operation involves removing the colon and the rectum, but the anus is left. An artificial rectum, called a pouch, is made out of the lower end of the small intestine (ileum). The pouch is joined to the anus so bowel actions can be controlled in the normal way. The pouch stores the faeces until the person goes to the lavatory in the usual way. Usually this operation is done in two stages.

Stage 1

The colon and rectum are removed and the pouch is made and joined to the anus. Because the operation is quite complicated, it is sometimes necessary to allow the new pouch time to "rest" while it heals. This is done by creating a temporary ileostomy above the pouch which means that stools have to be collected in a bag which is worn outside the body, on the abdomen (tummy). The bag will need to be emptied at certain times.

The average stay in hospital is around 10 - 12 days after the operation. Return to light work is usually possible between four to six weeks postoperatively.

Stage 2

After at least 2 months, when the pouch has healed, the ileostomy is closed by a second operation so that stools are again passed from the anus in the usual way.

With this operation the average stay in hospital is a bit shorter (4 - 6 days).

Sometimes it is possible to do the operation without creating a temporary ileostomy. This should be discussed with the surgeon.

Sometimes mistakes occur in the DNA code so that genes carry wrong information. FAP occurs when the genetic recipe is wrong.

For example, consider a recipe. For a ham sandwich you need bread, butter and ham. If when writing the recipe the H in ham was changed to a J the end result would be a jam sandwich. Just a minor change but the end result is very different.

When the FAP gene carries the correct information, the colon is protected from developing polyps. When the FAP gene is faulty (so that incorrect information is carried) this protection is lost and the colon develops many polyps.

What is the chance of inheriting FAP?

FAP is usually inherited from a parent who has the condition. Each child, boy or girl, born to a person with FAP has a 50:50 chance of inheriting the gene that causes it. A 50:50 chance is the same as the chance of getting a head or a tail when you toss a coin. This is known as an autosomal dominant mode of inheritance.

If a person has not inherited the gene that causes FAP then that person's children will not be at any increased risk of getting polyposis.

Sometimes a person may be affected even though both parents have normal copies of chromosome 5. This is because mistakes can occasionally occur in the DNA as the chromosome is transmitted from parent to child.

In cases where this appears to have happened it should be confirmed. Parents, brothers and sisters should be screened. The children of a new person with FAP will have a 50:50 chance of inheriting the affected gene.

How would you know if you had FAP?

You might not. In the early stages there may be no symptoms. Because early diagnosis makes such a difference to people with FAP, anyone who thinks they might be at risk should not wait for symptoms to develop, but should seek advice from their G.P.

There are 2 ways of determining if a person has FAP:

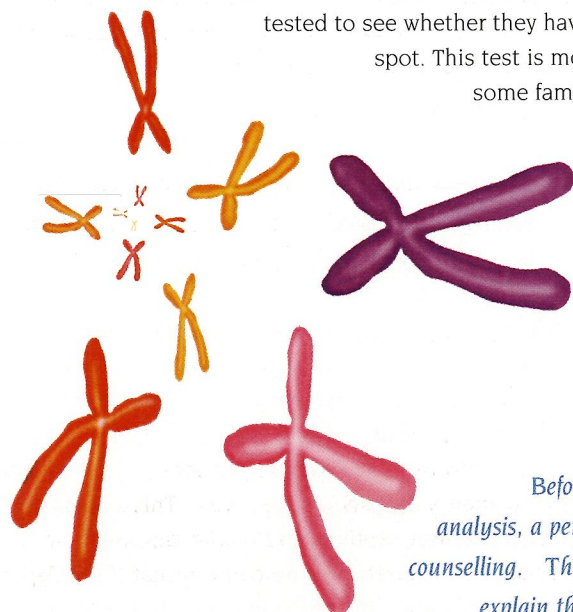
- i. DNA analysis
- ii. Bowel screening

DNA analysis

There have been many developments in this area.

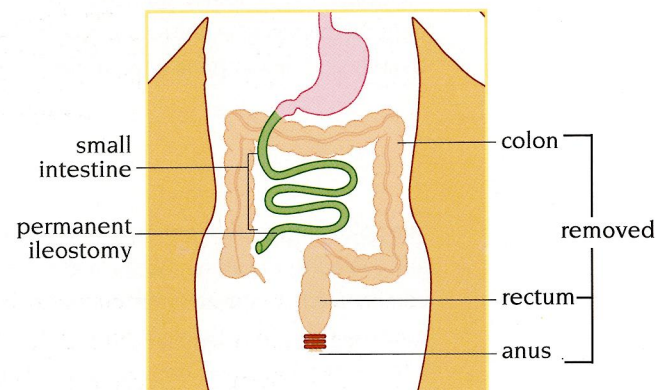
Now that it is known that the gene responsible for FAP is on chromosome 5, it is possible to take a sample of blood, extract DNA from the sample and then test it to see if the affected gene is present or not.

The fault in the gene varies from family to family so it is necessary to find the exact faulty spot in each family. Once it has been found other individuals in the family can be tested to see whether they have got the same faulty spot. This test is more difficult to do for some families and, at the moment, does not work for everyone.



Before blood is taken for DNA analysis, a person will be offered genetic counselling. The counsellor will be able to explain the situation in more detail.

The waste products will come out of the ileostomy into a discrete bag that is securely stuck onto the skin of the abdomen and which is worn under the clothes. With an ileostomy it is not possible to control when it will act but it is possible to control the emptying or changing of the bag. The stoma care nurse will provide support and education.



Life after the operation

After the operation it is usually necessary to stay in hospital for about 2 weeks. Return to normal activities can begin after about 6 to 8 weeks although strenuous activity should be avoided for about 3 months. The majority of individuals with an ileostomy will lead a normal life. Activities such as swimming do not need to be avoided. Some people find it best to avoid certain foods, especially those that are fibrous and pithy (for example oranges) because they can lead to obstruction of the small bowel.

Follow up after surgery

Once things have settled down it is usual to be seen in the outpatient department on a yearly basis.

Life after the operation

perhaps be avoided for about 3 months. Afterwards most people will go to the toilet 3 or 4 times a day and the stools (or faeces) may be softer than before. If a person finds that they need to go more often, tablets called loperamide (Imodium) can be taken. These tablets slow down the bowel and are not addictive.

Some people find that certain types of food upset their bowel and are best avoided. Others eat whatever they want when they want to. People differ greatly but beer, spicy foods, raw vegetables and fruit are often mentioned as causing loose bowel motions. Each person has to find out what suits them.

Follow up after surgery

People who have had a colectomy with ileorectal anastomosis will still have their rectum. Polyps may continue to develop in the rectum and it is important that the rectum is examined regularly which is done in the outpatient department every six months.

2

Total proctocolectomy and permanent ileostomy

This type of operation is now rarely done for polyposis. It involves removing the whole of the colon, rectum and anus. Because the anus has been removed it is not possible to control the bowel in the usual way.

The end of the small bowel (called the ileum) is brought out onto the abdomen where it protrudes about an inch. This is called an ileostomy. It is stitched into place so there is no need to worry about it falling back inside.

Suppose there is a suspected fault in a train and it is known that one of the bolts might be missing from under the back seat, on the left, in the fifteenth carriage. An engineer would soon be able to tell if there was a fault or not. This is like checking for a fault in a gene, in a family where the position of the fault has already been identified. But suppose nobody knows where the fault is in the train. In this case every carriage of the train would have to be examined thoroughly in order to find it. This is what has to be done when the faulty gene in a family has not yet been identified.

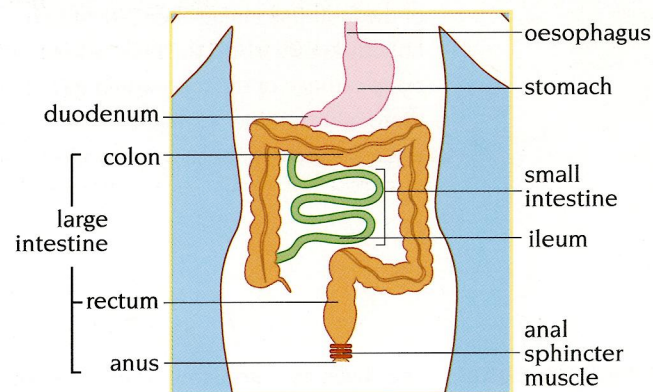
Bowel screening

The check-up is straightforward. The doctor will ask questions about general health, feel the tummy and carry out a test called a sigmoidoscopy. A sigmoidoscopy involves passing a small telescope into the bottom so that the doctor can have a look to see if there are any polyps. Some people find the examination a bit embarrassing and uncomfortable but it should not hurt.

Polyps can appear in people at different ages and if nothing is seen during an examination the person may be invited to take part in a screening programme.

If during the examination, the doctor sees polyps a small sample (called a biopsy) will be taken. This is not painful and cannot usually be felt. The sample is sent to the laboratory for analysis to confirm that the polyp is an adenoma.

The gastro-intestinal tract



The intestine is divided into three parts:

1. The first part is the stomach. When food is swallowed it goes into the stomach where it is churned around and mixed with gastric juice.
2. The second part is the small intestine or small bowel. The blended food passes from the stomach into the small bowel where nutrients from food are absorbed.
3. The third part is called the large intestine. In the diagram you will see that the large intestine is itself divided into three parts, the colon, the rectum and the anus. The main function of the colon is to absorb fluid. The remains are passed into the rectum which acts like a storage area before these waste products (faeces) are passed from the anus when going to the lavatory.

How is FAP treated?

There are too many polyps to remove one by one. Three types of operation are used:

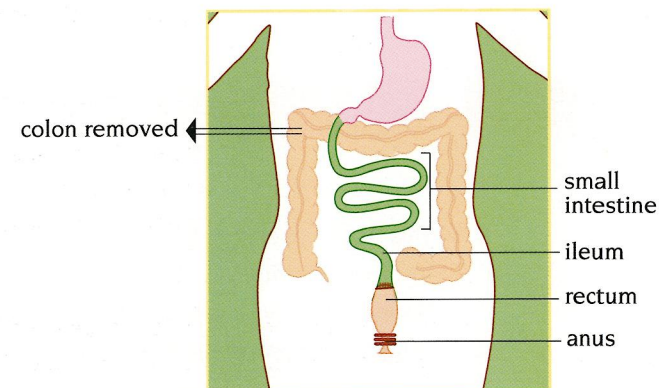
- 1 Colectomy with ileo rectal anastomosis (IRA)
- 2 Total proctocolectomy with permanent ileostomy
- 3 Restorative proctocolectomy (Pouch)

Following an examination the Surgeon will discuss with the patient which type of operation should be done. These operations are described in more detail below.

1

Colectomy with ileorectal anastomosis (IRA)

This operation involves removing all of the colon. The end of the small bowel (the ileum) is then joined (anastomosed) to the top of the rectum. That is why it is called an ileo rectal anastomosis or IRA for short.



It may be necessary to stay in hospital for 6 - 8 days. Most people will get back to normal life in about six weeks. Strenuous exercise, such as lifting heavy weights, should