

Familial Adenomatous Polyposis (FAP)

Key Points

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- FAP is a rare condition associated with multiple bowel polyps or outgrowths from the lining of the large bowel.
- There are different types of bowel polyps that can occur in the large bowel or colon. The type of bowel polyp a person has is determined by looking at a sample (biopsy) of the polyp under a microscope.
- People with FAP develop adenomatous bowel polyps, hence the name of this condition.
- People with FAP usually develop multiple large bowel polyps before the age of 20 years. Adenomas in the general population do not usually occur until the age 40 – 50 years and then only in small numbers.
- Without treatment, individuals with FAP will inevitably develop bowel cancer in mid life.
- Cancer occurring in individuals with FAP accounts for less than 1% of all bowel cancer.
- Bowel surgery prevents individuals with FAP from developing bowel cancer. This surgery is normally recommended in the later teenage years.
- Yearly bowel screening is required for the part of the large bowel that remains to prevent cancers developing.
- People with FAP have up to a 4 - 5% risk of developing upper gastrointestinal cancer in their lifetime and should have regular upper gastrointestinal endoscopy from the age of 25- 30 years.
- FAP is caused by an alteration in the APC gene. The alteration in the gene means that it cannot do its job properly.
- Up to 30% of people with FAP do not have a family history of the condition, i.e. they are the first person in their family to develop an alteration in the APC gene.
- Once present, FAP is a dominantly inherited condition. This means that if you have FAP each of your children has a 1 in 2 (50%) chance of developing FAP.
- Genetic testing to try and identify an abnormality in the APC gene is available for families with this syndrome.
- The aim of the NZFGICS is to reduce the number of cancers occurring in families by facilitating the required bowel screening for registered families.

What is FAP?

FAP is an inherited condition which causes people to develop multiple (usually at least a 100) adenomatous bowel polyps. Bowel cancer develops from adenomatous bowel polyps over a period of time. Individuals with FAP will inevitably develop bowel cancer without surgery to remove the majority of their large bowel.

What is the risk of developing bowel cancer in FAP?

Almost all individuals with FAP will develop bowel cancers by their late thirties and forties. This differs from other people who tend to be over the age of 50 years when they develop bowel cancer.

What is attenuated FAP (AFAP)?

Some families have attenuated FAP (AFAP). This is a later onset type of FAP. Individuals with AFAP develop fewer polyps at a slightly older age than those with typical FAP. Although people with AFAP tend to develop colon cancer at a later age, they still have a very high lifetime risk of colon cancer.

What are the bowel symptoms of FAP?

In FAP polyps usually start to develop throughout the colon, before the age of 20 years. Adults may have hundreds to thousands of polyps. There may be no symptoms to alert you that you have bowel polyps. The major concern is that the polyps will become cancerous.

Does FAP have other effects on the body?

FAP can have other effects on the body as well as causing bowel polyps. Polyps are commonly seen in the stomach but these are a different type of polyp and are usually harmless. Some people with FAP get skin cysts or harmless bumps on their bones, or Freckle-like spots on the inside lining of the eye, which we call CHRPE, can also occur but these do not affect vision in any way.

A small number of people with FAP will develop benign growths in their abdomen called desmoids tumours. Occasionally they cause problems but they do not become cancerous.

There is no need to have regular checks for desmoids but if you have any skin or bone lumps or unexplained symptoms, you should discuss them with your General Practitioner (GP) or let your specialist know at your next visit.

What causes FAP?

In FAP, part of a gene called APC is altered, so it cannot do its job properly. People with FAP have one working copy and one altered copy of the APC gene. The altered copy of the gene is responsible for the development of the large bowel adenomatous polyps.

Who is at risk of FAP?

When you have FAP each of your children has a 1 in 2 (or 50%) chance of inheriting it.

What does the genetic test result mean for my family?

The gene involved in FAP (called APC) is like a very long instruction sentence. A spelling mistake or alteration in any part of this instruction can cause FAP. Each family with FAP will have a different alteration but the individuals within the family will have the same alteration which is called a mutation. We can look in a blood sample from someone who has FAP to try to find the alteration in a family.

If we do find a genetic mutation we can test other family members to see if they have inherited it. We recommend that children have a genetic test around the age of 12-14 years to see if they have inherited the altered gene.

If they have not inherited the altered FAP gene, they do not need FAP related bowel checks but if they develop bowel symptoms then like anybody else they need to see their GP.

Individuals who have not inherited the altered FAP gene will not be able to pass the altered gene on to their children in the future.

If an individual has inherited the altered FAP gene it is important that they have bowel checks every year starting from the age of 14-15 years.

What if no spelling mistake is found?

If we don't find a gene alteration we won't be able to offer a genetic test to other relatives but they will still need bowel checks. This is generally advised from teenage years but we will discuss the best screening plan for your family with you.

Most people with FAP develop polyps, usually in the teenage years. However, if genetic testing is not available and an individual at risk of having FAP has been having regular colonoscopy and no bowel polyp have been identified by the age of 50 years it is unlikely that they have FAP. In this situation the regular bowel checks would stop. However, even when the bowel checks have stopped if an individual develops new bowel symptoms they may still need to be investigated because they can still get bowel problems like anyone else in the general population,

What bowel screening checks do I need and how often do I need it?

Most people who have FAP begin to develop polyps in their early teens. People at risk of FAP should start bowel checks between 14 and 15 years of age. These should be repeated every year. It is important to remember that it is not safe to simply wait for symptoms to develop. It is vital that children at risk of FAP have regular bowel checks even if they do not have symptoms.

Do I need other checks?

Most individuals with FAP develop polyps in the first part of the small bowel (duodenum) by the time they are in their seventies but only a small percentage (approximately 5%) develop duodenal cancer.

A tube test examination of the stomach and first part of the small bowel (gastroscopy) is recommended from the age of 25-30 years to identify those individuals with FAP who may develop troublesome duodenal polyps. The interval between procedures is determined by the number and type of polyps found and can be anywhere between 1 -5 years.

What is the treatment?

Once large bowel polyps are found individuals with FAP will be advised to have surgery to remove the large bowel.

Surgery is generally planned towards the end of the teenage years, once schooling has been completed.

There are 2 main types of operation:

- Colectomy and ileo-rectal anastomosis (IRA) involves removing the large bowel and attaching the small bowel to the rectum or bottom part of the large bowel. The remaining rectum can still develop polyps so this must be examined every twelve months to identify and remove these polyps in order to prevent a cancer starting there. An individual who has had a colectomy and IRA may need further surgery if an unmanageable number of rectal polyps develop. This surgery involves removing the remaining bottom part of the bowel and creating a new 'back passage' as described below.
- Sometimes at the age when the initial surgery is being planned more extensive polyps have already developed in the rectum and colon and therefore the planned surgery involves removing the whole rectum and colon. This is called a pan procto-colectomy. A new "back passage" or ileal anal pouch is created. The small bowel is fashioned into a new rectum "pouch" and connected to the anus. This means you can pass a motion normally through your anus. Following pouch surgery regular endoscope checks for polyps are required.
- A pan procto-colectomy and ileo anal pouch may also be planned as the initial surgery if desmoid disease is present in your family and if it seems likely that desmoid disease will be present at the time of surgery. This type of surgery is advised in this situation because desmoids can grow after surgery and make further surgery more difficult.
- An ileostomy is when the large bowel is removed and the end of the small bowel is brought to the surface and the waste material is collected in a disposable bag. Quite often after pouch surgery you do need an ileostomy for a short time while the pouch heals. Your surgeon is best able to advise you as to further details of these

Does diet matter?

At the moment there is no known effect of diet on those with FAP. We recommend a healthy diet and lifestyle. Healthy foods include fruit, vegetables, cereals, bread (whole meal), pasta, rice, olive oil and non-fat dairy food such as yoghurt.

Possible harmful foods are excessive amounts of animal fat, charred meat, beer and cigarettes.

For more information visit <https://www.health.govt.nz/resource/eating-healthy-adult-new-zealandersng%C4%81-kai-t%C5%8Dtika-ma-te-hunga-pakeke-o-aotearoa>

Is there medication available?

There is evidence that the different types of Non Steroidal Anti-inflammatory Drugs (NSAIDs) can decrease both the number and size of adenomatous polyps in the large and small bowel. These drugs may at times be used to control polyp growth while waiting for surgery. They are not routinely used because NSAIDs have a range of side effects and the benefits of using these drugs has to be carefully weighed up against the potential risk associated with their use.