

# Familial Adenomatous Polyposis (FAP)

## What is FAP?

FAP stands for Familial Adenomatous Polyposis. Polyposis means lots of polyps, adenomatous is the type of polyp and familial means it runs in families.

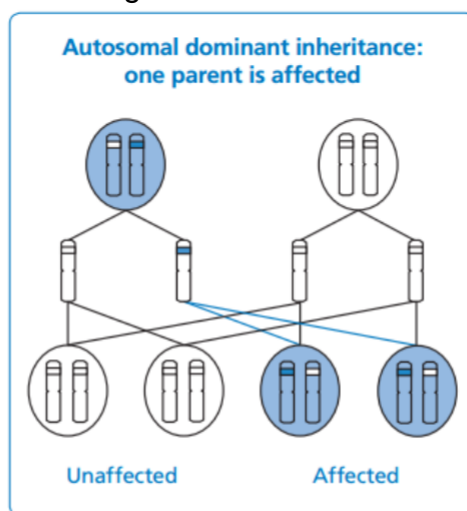
Individuals with FAP have an increased chance of developing cancer. It causes multiple polyps (small growths) to develop in the bowel. The polyps usually begin to appear in adolescence, but the age at which they develop can be variable. The polyps themselves are not dangerous, but if they are left untreated one or more will turn into cancer. The condition affects both men and women.

## What causes FAP?

FAP is a genetic condition and is caused by a change in a gene known as APC. Genes come in pairs because we get one copy from each parent. In FAP, one copy of the APC gene has a fault in it and the other copy is normal. FAP is called a dominant condition because you only need one faulty copy of the gene to have the condition.

## The inheritance of FAP

FAP is usually passed from parent to child (of either sex). Some people with FAP are the first person diagnosed with the condition in their family. This may be because the fault in the gene occurred for the first time in the sperm or egg that made them, or it may be that FAP was not recognised in previous generations of the family. Each time an individual with FAP has a child, the child has a 50% chance of having FAP and a 50% chance of not having FAP.



## Genetic testing for FAP

We can look for the exact APC gene fault which has caused FAP if we have a blood sample from a person who has the condition. In different families, there are different mistakes in the gene. If we know the mistake in the family, we can look just for that. If you are the first person to have FAP or we don't know the mistake in your family, we can look through the whole gene for a mistake. We can't always find the mistake, but we do in most people (80%) of those with FAP.

## Family members of a person with FAP

It is helpful to know if relatives of a person with FAP will develop the condition, so they can have the right medical management. If the exact gene fault is known in a family, then we can offer a blood test to relatives to see if they have inherited FAP. The test is available from 10 years of age because polyps usually start growing around puberty.

Alternatively (or if the gene fault has not been identified) a camera examination of the bowel (colonoscopy) can be carried out to look for bowel polyps. The frequency of screening depends on whether polyps are found in the bowel.

## Management and treatment for FAP

People with FAP need to have a camera examination (colonoscopy) of the bowel every 1-3 years to monitor the number and size of polyps. Initially, polyps can be removed during this examination, but as their number increases this becomes too difficult.

Eventually, planned surgery to remove the large bowel is required to prevent cancer. Sometimes this means using a temporary bag (stoma), but this is rarely permanent. Normal bowel function is often regained after surgery.

Polyps can also grow in the stomach and the duodenum (the exit from the stomach). These polyps are much less likely to turn into cancer. These polyps need to be checked every 3-5 years from the age of 30. This is done by a camera examination down the throat.

## Other features of FAP

There are a few other features that are associated with FAP, such as extra spots of pigment at the back of the eye and bony lumps on the jaw, but these usually go unnoticed and do not cause any problems.

There are some other medical problems associated with FAP. These are rare and do not happen often. These include non-cancerous soft tissue tumours, desmoid tumours (specific non-cancerous tumour seen in connective tissue), and associated cancers such as thyroid cancers. If you have questions about these please talk to your genetics clinician.

## Attenuated FAP

Some families with FAP have fewer polyps and these develop later in life. This is called attenuated FAP. It is inherited in the same way as classical FAP but means less frequent screening is necessary, and it can often start at a slightly older age.

## Additional Support

There are two main UK patient Support groups:

The FAP Gene Support Group: <http://www.fapgene.com/> who also run a closed facebook page.

Polypeople attached to St Marks in London and for all polyposis conditions including FAP:  
[www.polypeople.online](http://www.polypeople.online)

## For more information

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

### **Manchester Centre for Genomic Medicine**

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Department staffed Monday–Friday, 9.00 am to 5.00 pm.

Website: [www.mangen.co.uk](http://www.mangen.co.uk)

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Seen in clinic by (doctor): \_\_\_\_\_

And (Genetic Counsellor): \_\_\_\_\_

Telephone number: \_\_\_\_\_

Family reference number: \_\_\_\_\_