

About Hereditary non-polyposis colorectal cancer (HNPCC):

- HNPCC is an inherited tendency to develop colorectal (bowel) cancer. Families with HNPCC also have an increased risk of certain other cancers, including cancer of the uterus, ovaries, stomach, urinary tract, small bowel and bile ducts.
- HNPCC often occurs at a younger age than non-inherited colorectal cancer. Unlike other forms of bowel cancer, HNPCC can occur with only a small number of polyps present in the bowel, or when no polyps at all are present.
- Early diagnosis is important for detecting cancer as it increases the chances of successful treatment. People who are known to be at risk of HNPCC are offered regular bowel screening (colonoscopy), and women are also offered regular pelvic and uterus screening.
- HNPCC is caused by an inherited change (a mutation) in any one of several different genes. People with a HNPCC gene mutation have a 50 per cent chance of passing it on to each of their children. In some cases, there is no family history of the disease, but these patients can still pass the mutation on to their children. Most people who inherit an HNPCC gene mutation will develop cancer at some point in their lives.
- Once a HNPCC gene mutation has been identified in an affected person, a genetic test may be offered to other family members who might have inherited the condition. However, some of the genetic changes that cause HNPCC have not yet been discovered, and so not all affected families can be offered this type of test.

Find out more

To find out more about HNPCC, and for information and support contact:

Colon Cancer Concern
08708 506050
www.coloncancer.org

or Cancer BACUP.
0808 800 1234
www.cancerbacup.org.uk



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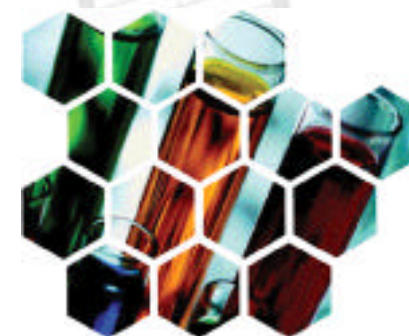
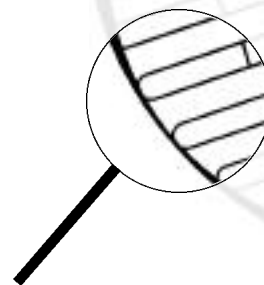
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Getting a diagnosis of...



Hereditary non-polyposis colorectal cancer (HNPCC)



HNPCC

Who should have a genetic test for Hereditary Non-polyposis Colorectal Cancer (HNPCC)?

It is important to understand that while (as best as we know today) many cancers may be influenced to some extent by heritable genetic variants, most cancers aren't hereditary as such. More often, altered genes may arise from a new, non-inherited gene mutation during the course of a person's life, and therefore cannot be predicted from the family history.

Cancer that is hereditary is caused by an altered gene that has been passed from generation to generation. One such cancer is Hereditary Non-polyposis Colorectal Cancer (HNPCC), which is estimated to account for about 2-4% of all cancers of the large bowel. While it is the most common hereditary colon cancer and may be caused by an abnormality in any one of at least five genes, the vast majority of colon cancers still arise without such a hereditary cause. Therefore, the test for HNP is not recommended for everyone. It makes sense, only in families where the presence of this predisposition can be traced through a family tree.

Approximately 60-70% of HNPCC patients have mutations within either one of two genes; one on chromosome 3 and one on chromosome 2. These genes code for proteins that can repair errors occurring during the process of copying DNA as cells are dividing to form new cells. Those with HNPCC have inherited one faulty copy of the gene; if the second gene copy then becomes damaged, any errors occurring in the DNA in that cell cannot be corrected. An accumulation of errors within the DNA leads to the development of a cancerous cell and eventually a tumour.

People who are carriers of HNPCC (i.e. those who have inherited the genetic mutation) have an increased risk of early onset colorectal cancer as well as other cancers. If cancers are detected at an early stage, they can be cured by surgery in 90% of patients. However, if they are detected only at a later stage, this survival rate drops. Therefore, once someone has been identified as having as a carrier of HNPCC, he or she needs to have regular and thorough checkups performed.

The lifetime chance that a person who carries a mutation (in one of the genes that cause HNPCC) will develop bowel cancer is between 65% and 90%. Women mutation carriers are less likely than men to develop bowel cancer.



Annabel* lives in Surrey, where she works in middle management. Last year, she discovered that she has inherited a tendency to develop a condition called HNPCC (hereditary non-polyposis colorectal cancer), as well as certain other types of cancer.

Annabel was offered a test for HNPCC because several members of her family had developed bowel, stomach or womb cancer at an unusually young age. The test results confirmed she too had inherited an increased risk of cancer:

'My brother and I were tested together and received the results on the same day. I knew immediately that he was not carrying the gene because he returned so quickly after seeing the consultant. When I went in I felt sure I had the gene, but when I was told, although I expected it, it was such a shock.'

Annabel was told that she has inherited a genetic change that means she has a 60-90 per cent lifetime risk of developing certain types of cancer. She now has regular check-ups to pick up any early signs of the disease:

'I was told that there's a greater risk that I could get cancer at some stage during my life, and it was advised I had certain tests - I've got to have a test for bowel cancer every two years... pelvic screening, a mammogram and a smear test every year... you need to be more aware of health issues - you don't want to be going to the doctor every five minutes, but you do need to be a lot more vigilant in your approach.'

Annabel says that getting the test results have affected her life in several ways:

'I think it changes everything...if I have a child, I might pass on that gene to my child, and the other side is the insurance... my father has had problems and I'd already taken out insurance, critical illness cover anyway, because I was aware that there was a possibility that I may have to have these tests - so it can have a huge effect.'

As well as getting health insurance, Annabel was also concerned that the test result might affect her employment prospects:

'That's one reason why I don't tend to tell people...at the time I was working, and I had to inform my immediate boss because I had to take time off for tests... if I had an interview for a new job and I told them about the gene, they may think twice about employing me because I may be a risk.'

Despite these difficulties, Annabel says she is glad she decided to have a genetic test for HNPCC, rather than just going for screening tests:

'I know some people might say they'd rather not know whether or not they carry the gene, but I think it's better to know and then have the relevant tests, it could save your life.'

* Not her real name