

BOWEL CANCER and GENETICS

Is bowel cancer inherited?

Bowel cancer is the common name for a carcinoma (*or adenocarcinoma*) of the large bowel (*the colon and rectum*). This type of cancer is not usually inherited. An inherited susceptibility for bowel cancer is estimated to exist in about 10% of people who develop this disease. This susceptibility is often a single altered gene. It is true to say that all cancer cells contain some genes that have malfunctioned, this allows the cells to behave in an abnormal way and grow as a cancer, yet most of the gene faults (*or mutations*) within a cell are acquired after birth rather than due to an inherited gene abnormality.

What causes these gene changes?

Genes are made from a chemical called DNA (*deoxyribonucleic acid*). This DNA must copy itself each time a cell divides. Occasionally this copying mechanism makes a mistake and this may have a detrimental effect on a gene. These errors occur in all of us but not everyone will inevitably develop cancer. A cell also has repair mechanisms to correct these DNA errors but these repair mechanisms are not infallible. Things like radiation and some chemicals (*for example in cigarette smoke*) can damage DNA. Usually the exact events that cause DNA damage in an individual cell are not known. This DNA damage does tend to accumulate with age, so most bowel cancers (*with some important exceptions*) occur in people over the age of 50.

How do I know if I have a gene that can cause bowel cancer?

The most helpful clue is to document the family history going back to grandparents on both sides. This history should include all uncles, aunts and cousins. Gene testing remains a difficult undertaking but it is available to those families with a strong family history. This testing is available at no cost to the patient in the public hospital system. An example of a strong family history would be if three or more people on the same side of the family have had bowel cancer.

Other clues listed below are also taken into account in deciding who is eligible for gene testing.

- Age of onset of cancer
- Other cancers that may have occurred in the family
- How many polyps are found in the colon
- Location of the cancer (*right or left side of the colon*)
- Pathology of the tumour
- Specialised tests of the tumour DNA itself
- Staining for some of the hereditary bowel cancer gene proteins within the cancer cells

Can I get a bowel cancer gene test?

Testing is only possible after genetic counselling has taken place. Gene testing will generally start with a blood sample from an affected person within a family. Testing all the known bowel cancer genes is simply not possible, so it is important to try and determine which bowel cancer gene may be the most likely one responsible in each individual family.

How many bowel cancer related genes are there?

Nearly a dozen or so bowel cancer genes have been found. The first inherited bowel cancer gene that was discovered was the APC gene. This gene was discovered in 1991. It causes a medical disorder called Familial Adenomatous Polyposis (*FAP*). The next inherited bowel cancer syndrome identified was called Hereditary Non Polyposis Colon Cancer (*HNPCC*) or Lynch syndrome. There are several genes that can cause HNPCC. Some genes yet to be discovered are likely to cause clustering of bowel cancer in still other families. Unfortunately there are many families with bowel cancer in which no responsible gene can be identified with current gene testing techniques.

I have a family history of bowel cancer and want to find out more Information

Your own doctor may be able to refer you to a family cancer clinic or the nearest genetics clinic in a public hospital. The cancer information telephone line (*call 131 120 in Australia*) will also be able to tell you where the nearest cancer genetic service is located.

The National Health and Medical Research Council have produced a publication called, "Guidelines for the prevention, early detection and management of colorectal cancers: A guide for patients, their families and friends". This booklet has a chapter on bowel cancer in families. The booklet is available free on the internet at: www.nhmrc.health.gov.au/publicat/cp-home.htm. It has 85 pages and so it might be preferable to purchase a copy from the AusInfo Government bookshop by calling 132 447. The booklet costs \$10.80, including postage.

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