Bowel (colorectal) cancer is the second most common cause of death due to cancer in Australia.

- The lifetime risk to age 85 in the general population is 1 in 10 for men and 1 in 14 for women.
- In 2004 there were 12,973 new cases and 4,077 deaths.

The causes of bowel cancer are complex and involve interactions between environmental and genetic factors.

Cancer develops as the result of a multi-step process involving genetic mutations in cells lining the bowel wall. Most bowel cancers arise from adenomatous polyps.

Prevention

The following healthy lifestyle recommendations may reduce risk of bowel cancer and should be recommended to people of all ages:

- exercise regularly
- maintain a healthy weight including
  - limiting energy intake
  - reducing dietary fat (<25% of calories as fat);
  - consume poorly soluble cereal fibres
  - eat vegetables and fruit
- avoid or limit alcohol consumption
- do not smoke, (or quit smoking)

Screening for bowel cancer for everyone from the age of 50

Research shows that about 90% of bowel cancer can be cured if detected at the earliest stage.

Randomised controlled trials have shown that population screening for bowel cancer using faecal occult blood testing (FOBT) is effective in reducing mortality from this disease.

FOBT is recommended at least every 2 years for all people over the age of 50. The Australian Government has committed to phase in a National Bowel Cancer Screening Program. Further information about the National Bowel Cancer Screening Program may be obtained by visiting the website www.cancerscreening.gov.au

A number of case-control studies have shown that sigmoidoscopic screening reduces mortality from bowel cancer but randomised controlled trials have not yet been completed.

A family history of bowel cancer

About 15-20% of people who develop bowel cancer have a first-degree relative (parent, sibling, child) also affected by the disease.

More than one blood relative in the family with bowel cancer can occur:
- just by chance (usually the reason),
- because family members have environmental and lifestyle factors in common (eg. some aspects of their diet), or
- because of an inherited genetic predisposition to bowel cancer (relatively uncommon).

Identifying individuals at increased risk of bowel cancer

Individuals with a family history of bowel and perhaps other cancers may be at increased risk of developing bowel cancer.

Family history of bowel cancer can be used to estimate the risk of bowel cancer. This may help to determine the type and frequency of bowel cancer screening. Referral to a familial cancer or genetic counselling service may be appropriate for some families (see overleaf).

Establishing a person’s family history of cancer

- Ask about all first- and second-degree blood relatives on both sides of the family (including those who are interstate or overseas).
- First-degree (1°) relatives: parents, siblings, children
- Second-degree (2°) relatives: grandparents, grandchildren, aunts, uncles, nieces, nephews
- Record the primary site and age at diagnosis of any cancer: people may have imprecise knowledge of cancer in their relatives. Death certificates are a good source of information.
- Regularly update the family history.

Bowel cancer known to involve a genetic predisposition

Familial Adenomatous Polyposis (FAP)

- A rare condition, usually due to a mutation in one of the two copies of a tumour suppressor gene called the **adenomatous polyposis coli (APC)** gene.
- Individuals with a mutated APC gene usually develop hundreds of adenomas throughout the colon and rectum that may appear in the teenage years or in early adult life. If left untreated, one or more of these adenomas will progress to cancer, often at an early age, so prophylactic surgery must be considered.
- Pathological lesions may occur outside the large bowel, such as upper GI cancer (especially of the duodenum), desmoid tumours and osteomas.
- Inheritance of a mutated APC gene follows an autosomal dominant pattern. Sometimes there is no family history because a new mutation has occurred around the time of conception. This happens in 20-30% of cases.
- Rarely, a condition similar to FAP is due to recessive inheritance of mutations in both copies of the **MUTYH** gene.

Hereditary Non-Polyposis Colorectal Cancer (HNPCC) – also known as Lynch Syndrome

- A rare condition due to an inherited mutation in a copy of one of a group of DNA mismatch repair (MMR) genes.
- Inheritance of a mutated MMR gene follows an autosomal dominant pattern. Sometimes there is no family history because some individuals with a mutated MMR gene will not develop cancer (or have not done so at the time the history is taken).

- Individuals most likely to have a mutated MMR gene copy are those from families with a strong history of colorectal cancer, characterised by: early age of onset (<50 years), and a tendency for proximal colonic malignancy or multiple colorectal cancers.
- Cancers occurring outside the large bowel may also be a feature. The most common of these is endometrial cancer, but the syndrome also includes cancers of the ovary, stomach, small bowel, renal pelvis or ureter, biliary tract, and brain.
- Cancers associated with HNPCC tend to show high levels of microsatellite instability and may lack immunohistochemical expression of MMR proteins in tumour tissue. Consideration of tumour testing should be given for all patients diagnosed with colorectal cancer aged 50 or under, or where there is a suggestive family history.

Familial cancer and genetic counselling services

(Phone 13 11 20 for contact details)

These services provide risk assessment, genetic counselling and, if appropriate, genetic testing for a causative mutation where there is a strong family history of cancer and/or where tumour testing suggests HNPCC.

- For both FAP and HNPCC, if a causative mutation can be identified in an affected individual, then blood relatives can be offered predictive genetic testing.
- Those found to have inherited a gene mutation that confers a high risk of developing bowel cancer can be offered individualised cancer screening and strategies for prevention.
- Blood relatives proven not to have inherited the family-specific mutation still have an average risk of developing bowel cancer based on their age and should follow recommendations for population screening. However, they can be spared the intensive screening needed by someone who has may have the mutation.
- Sometimes a causative mutation cannot be found in a person with FAP or HNPCC. In this case, it can’t be presumed that a mutation is not present. Therefore first-degree relatives should be considered to be at 50% (or 1 in 2) risk of having inherited a mutation and participate in a screening and prevention program according to appropriate guidelines. (See Category 3 Potentially high risk).

Hereditary Bowel Cancer Registers

Assist familial cancer and genetic counselling services and other clinicians to identify, manage, support and follow-up people at high risk of bowel cancer due to a family history. Contact details are available from The Cancer Council Helpline: Phone 131120.
INFORMATION FOR CONSUMERS

BOWEL CANCER AND FAMILY HISTORY

What is cancer?
Cancer occurs when the cells in the body become abnormal and grow out of control.

What is bowel cancer?
The bowel is made up of two sections: the small bowel, where food is absorbed into the body and the large bowel, where only salts and water are absorbed. The large bowel has two parts: the colon and the rectum. The rectum leads to the outside of the body via the anus (back passage).

Bowel cancer generally refers to cancer of the large bowel (made up of the colon and rectum). Bowel cancer is therefore also known as colorectal cancer.

How common is bowel cancer?
Australia has one of the highest rates of bowel cancer in the world. It is the second most common cause of death from cancer in Australia.

In Australia, about 1 in 10 men and 1 in 14 women will develop bowel cancer before the age of 85.

The older you are, the greater your chance of developing bowel cancer. While it affects mainly people over 50, bowel cancer can occur at any age.

What about lifestyle factors?
It is recommended that people of all ages follow healthy lifestyle recommendations including:

- exercise regularly
- maintain a healthy weight
- do not smoke (or quit smoking)
- avoid or limit alcohol consumption

What about a family history of bowel cancer?
Bowel cancer is common. Many people have someone in their family who has (or had) bowel cancer. More than one person in the family with bowel cancer can occur:

- just by chance (usually the reason),
- because family members have environmental and lifestyle influences in common (eg some types of food they eat), or
- because of an inherited gene fault which causes a high risk of bowel cancer (relatively uncommon).

How can knowing about my family history of bowel cancer help?
If you have a family history of bowel cancer (no matter what your age) it is very important to talk with your doctor about this. It is also important to update your doctor about new cancers in blood relatives when you find out about them.

Recording the people in your family with bowel cancer and the ages at which they were diagnosed can help your doctor estimate your chance of developing bowel cancer.

1. Most people have around the average chance for the Australian population. This chance depends on your age.
2. Some people have a moderately increased chance of developing bowel cancer because of their family history. This includes people who have:
   - one first-degree relative (parent, brother, sister or child) with bowel cancer diagnosed before they were 55 years old
   - two close blood relatives on the same side of the family diagnosed with bowel cancer. In this case your doctor may need to ask further questions about you and your family history to estimate your chance of developing bowel cancer
3. A few people may have a potentially high chance of developing bowel cancer. These are people with:
   - more than two close blood relatives on the same side of the family diagnosed with bowel cancer
   - a family history of an inherited bowel condition such as familial adenomatous polyposis (FAP) or hereditary non-polyposis colorectal cancer (HNPPC) or other rare conditions. HNPPC is also known as Lynch syndrome.

In some situations genetic testing is available for people with a strong family history of bowel cancer so they can find out if they have inherited a faulty gene that places them at high risk of bowel cancer. If they have not inherited the faulty gene causing bowel cancer in their family, their own risk of bowel cancer is the same as the average chance.

Whichever of the above groups you are in, there are screening tests that can pick up bowel conditions at an early stage before there are any outward signs. Your doctor can work out which group you are in, advise you about how to prevent bowel cancer, the types of screening tests and how often to have them.

FOBT is recommended at least every 2 years for all people over the age of 50. A national bowel cancer screening program, using FOBT, is being implemented in Australia. Information on the national screening program is available from www.cancerscreening.gov.au or by contacting The Cancer Council Helpline (Phone 131120).

Sigmoidoscopy (rigid or flexible)
A test to examine the rectum and lower part of the colon (large bowel). A tube is inserted into the anus to view the lining of the bowel. Sigmoidoscopy can be done with a rigid or flexible instrument. Flexible instruments can reach further in the rectum and colon than rigid instruments and are more commonly used at present.

Colonoscopy
The doctor uses a longer flexible tube-like instrument called a colonoscope to examine the lining of the rectum and colon. Before the test, clear fluids are given and a special liquid medication is prescribed in order to empty the bowel in preparation for colonoscopy. The procedure is usually done under sedation. Any polyps (small growths) can be removed during this procedure, if they are seen.

CT Colonography
An imaging procedure using x-rays and computers to examine the large bowel.

Genetic testing
Laboratory testing of a blood sample to check for the presence of a gene fault associated with FAP or HNPPC. Such testing is available through familial cancer services. However, it may take weeks or months to complete.

FOR MORE INFORMATION...
Publications about bowel cancer prevention and screening, and information about services, are available from your State or Territory cancer organisation’s Cancer Council Helpline. Phone 13 11 20.

Family Health Trees
Contact your local genetic counselling service for a guide to drawing your family health tree. The Cancer Council Helpline has contact details.

Familial cancer and genetic counselling services
These services give people information about their chance of developing cancer based on their family history. Ways that might help reduce the chance of cancer or find it early are discussed. A doctor’s referral may be needed to attend.

Sometimes genetic testing is possible, but it is only done after the benefits and potential problems of testing for the person and their family have been discussed.

Hereditary Bowel Cancer Registers
Assist familial cancer and genetic counselling services and other clinicians to identify, manage, support and follow-up people at high risk of developing bowel cancer due to an inherited condition. For contact details phone The Cancer Council Helpline on 13 11 20.

Research shows about 90% of bowel cancer can be cured if picked up at the earliest stage.

What are the different types of tests?
Types of tests that might be done can include:

Faecal occult blood test (FOBT)
A test for traces of blood (which you might not be able to see) in the bowel motion. If blood is found, it does not mean that you have bowel cancer but other tests will have to be done to determine whether there is a problem.

The financial assistance of the Australian Government Department of Health and Ageing is acknowledged in the development of this publication.
# ASSESSING THE RISK OF BOWEL CANCER

## ASSESS THE FOLLOWING:

<table>
<thead>
<tr>
<th>PERSONAL HISTORY</th>
<th>FAMILY HISTORY</th>
<th>SYMPTOMS</th>
<th>FINDINGS</th>
</tr>
</thead>
<tbody>
<tr>
<td>• Bowel cancer</td>
<td>If there is a family history of bowel cancer, check for:</td>
<td>• Rectal bleeding</td>
<td>• Palpable abdominal mass</td>
</tr>
<tr>
<td>• Colorectal adenomas</td>
<td>• A history of multiple adenomas of the large bowel that may indicate the possibility of FAP.</td>
<td>• Symptoms of anaemia</td>
<td>• Enlarged liver</td>
</tr>
<tr>
<td>• Inflammatory bowel disease (IBD) of 8 years or more</td>
<td>• Other types of cancer that may indicate the possibility of HNPCC such as: endometrial, ovarian, stomach, small bowel, renal pelvis or ureter, biliary tract and brain.</td>
<td>• Change in bowel habit</td>
<td>• Palpable rectal mass (by digital rectal examination)</td>
</tr>
<tr>
<td></td>
<td>See notes on the front page about establishing family history.</td>
<td>• Abdominal pain</td>
<td>• Iron-deficiency anaemia</td>
</tr>
<tr>
<td></td>
<td></td>
<td>• Weight loss</td>
<td>• Positive FOBT</td>
</tr>
</tbody>
</table>

### NO PERSONAL HISTORY & NO SYMPTOMS OF BOWEL DISEASE

- Categorise according to family history

### PERSONAL HISTORY OF BOWEL CANCER, ADENOMA or IBD BUT NO SYMPTOMS

- Important to check family history as this may assist in planning the patient’s management.

### POSITIVE SYMPTOMS AND/OR FINDINGS

- Investigate
- Refer to appropriate specialist/s
- Important to check family history as this may assist in planning the patient’s management

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**NO FAMILY HISTORY**

**FAMILY HISTORY PRESENT - DETERMINE RISK CATEGORY (see below)**

If unsure about the significance of the family history, consult a familial cancer service for advice.

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Encourage patients to report early any rectal bleeding or persistent change in bowel habit.

**DRAW UP A FAMILY PEDIGREE**

These symptoms and findings suggest the possibility of Colorectal cancer and should be followed up accordingly.
### CATEGORY 1

**At or slightly above average risk**
Covers about 98% of the population

**Average risk**
- No personal history of bowel cancer, colorectal adenomas or chronic inflammatory bowel disease; and
- No confirmed close family history of bowel cancer.

**Risk - related to age (for both sexes)**

<table>
<thead>
<tr>
<th>Person's age</th>
<th>Risk in next 5 years</th>
<th>next 10yrs</th>
</tr>
</thead>
<tbody>
<tr>
<td>30 years</td>
<td>1 in 7,000</td>
<td>1 in 2,000</td>
</tr>
<tr>
<td>40 years</td>
<td>1 in 1,200</td>
<td>1 in 400</td>
</tr>
<tr>
<td>50 years</td>
<td>1 in 300</td>
<td>1 in 100</td>
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<tr>
<td>60 years</td>
<td>1 in 100</td>
<td>1 in 50</td>
</tr>
<tr>
<td>70 years</td>
<td>1 in 65</td>
<td>1 in 30</td>
</tr>
<tr>
<td>80 years</td>
<td>1 in 50</td>
<td>1 in 25</td>
</tr>
</tbody>
</table>

**Slightly above average risk**
- One 1° or 2° relative with bowel cancer diagnosed at age 55 or older.
- Two relatives diagnosed with bowel cancer at age 55 or older but on different sides of the family.

**Risk**
Up to 2 times greater than the average risk
(average risk according to age is listed in the table above).

People with affected relatives may have up to double the average risk, but most of this additional risk is expressed after the age of 60.

**Advise**
That their level of risk is at average or slightly above average, and that 90-95% of people in this group will never develop bowel cancer.

**Recommendations**
- Offer faecal occult blood testing (FOBT) for those aged 50 years or over.
- In addition, it is acceptable to offer sigmoidoscopy (preferably flexible) every 5 years.

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### CATEGORY 2

**Moderately increased risk**
Covers about 1 – 2% of the population

- One 1° relative with bowel cancer diagnosed before the age of 55 years (without potentially high risk features as in category 3).
- Two 1° or one 1° and one 2° relative/s on the same side of the family with bowel cancer diagnosed at any age (without potentially high risk features as in category 3).

**Recommendations**
- Offer colonoscopy every 5 years starting at age 50, or at an age 10 years younger than the age of first diagnosis of bowel cancer in the family, whichever comes first. Flexible sigmoidoscopy plus double contrast barium enema or CT colonography may be offered if colonoscopy is contraindicated for some reason.
- Consider offering FOBT in the intervening years. Patients should be informed that a positive test will require further investigation.

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### CATEGORY 3

**Potentially high risk**
Covers less than 1% of the population

- Three or more 1° relatives or a combination of 1° and 2° relatives on the same side of the family diagnosed with bowel cancer.
- Two or more 1° or 2° relatives on the same side of the family diagnosed with bowel cancer, plus any of the following high risk features:
  - Multiple bowel cancers in a family member
  - Bowel cancer before the age of 50
  - A family member who has/had an HNPCC-related cancer (endometrial, ovarian, stomach, small bowel, renal pelvis or ureter, biliary tract, brain cancer).
  - At least one 1° or 2° relative with a large number of adenomas throughout the large bowel (suspected FAP).
  - Member of a family in which a gene mutation that confers a high risk of bowel cancer has been identified.

**Risk**
It should be noted that not all individuals with the above family history will have a genetic susceptibility to bowel cancer (FAP or HNPCC). Without treatment, those with proven FAP have a lifetime risk of bowel cancer of almost 100%. For those with a mutation in a MMR gene, the lifetime risk is less, but may be up to 80% in some families. For family members shown not to have the mutation causing cancer in the family, risk is the same as that of the general population.

**Advise**
That their risk of bowel cancer is potentially high but surveillance and prophylactic measures are available (see below). Genetic testing is available for some to clarify risk.

**Recommendations**
- Consider referral to a familial cancer service for further risk assessment and possible genetic testing.
- Refer to a bowel cancer specialist to plan appropriate surveillance and management. This may include:
  - FAP: Flexible sigmoidoscopy yearly or second yearly starting from age 12-15 years until polyposis develops, then prophylactic surgery. If family genetic testing is inconclusive and no polyposis develops, sigmoidoscopy reduced to every 3 years after the age of 35, then change to population screening if examinations normal to age 55. Prophylactic surgery eg for large polyps or polyposis coli is appropriate for those with proven FAP.
  - HNPPC: Colonoscopy every one to two years from age 25, or five years earlier than the youngest diagnosis in the family (whichever comes first). FOBT may be offered in alternate years or to subjects unwilling to accept colonoscopy. There are options for surveillance at other sites, usually starting from age 25-35. Prophylactic surgery may be appropriate for some.

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**Randomised controlled trials have shown that population screening for bowel cancer using faecal occult blood testing (FOBT) is effective in reducing mortality from this disease.**
FOBT is recommended at least every 2 years for all people over the age of 50. A national bowel cancer screening program using FOBT is being implemented in Australia. For those not eligible for the national program, advice on access to FOBT is available from The Cancer Council Helpline (Phone 13 11 20).