

There are national guidelines for assessing the familial risk of breast cancer, ovarian cancer, and bowel cancer. These guidelines provide specific information about the assessment of individual risk on the basis of family history and can be obtained by calling the **Cancer Council Helpline 13 11 20** or on the Cancer Council Australia website at www.cancer.org.au/HealthProfessionals/clinicalguidelines/cancergenetics.htm

Familial cancer clinics

Familial cancer clinics are available in all capital cities and in large regional centres across Australia.

The services provided by these clinics include

- expert cancer risk assessment by a clinical geneticist
- genetic counselling
- information and advice about cancer prevention and early detection
- genetic testing (where useful and appropriate)

Genetic testing for familial cancer

In some cases, the Familial Cancer Clinic may offer a person genetic testing.

The aim of genetic testing is to identify the mutation responsible for the family's experience of cancer. However, genetic testing is not always possible or appropriate.

In many cases, the gene responsible for familial cancer is not known. There are also a number of important ethical, financial, and technical considerations.

Genetic testing should only be offered with appropriate pre- and post- test genetic counselling as the result may have significant implications for both the individual and other family members. The tests can be expensive, and samples may need to be sent interstate or even overseas for analysis.

For technical reasons, testing is usually initiated in a relative affected by cancer; testing an unaffected relative without the family's mutation having been identified is unlikely to provide useful information.

Genetic testing is now available for a number of familial cancers and familial cancer syndromes

- familial breast and/or ovarian cancer
- familial bowel and/or endometrial cancer (HNPCC)
- familial adenomatous polyposis (FAP)
- familial retinoblastoma
- Neurofibromatosis type 2 (NF2)
- Multiple Endocrine Neoplasia 2 (MEN2)
- Von Hippel-Lindau syndrome (VHL)
- Li-Fraumeni syndrome (LFS)
- Gorlin syndrome or Naevoid Basal Cell Carcinoma Syndrome (NBCCS)
- Hereditary diffuse gastric cancer.

Genetic testing has its limitations. It may not detect every mutation responsible for these familial cancer disorders. For this reason, genetic testing is not used to make a diagnosis of familial cancer in an affected patient. Instead, it is used to clarify the inheritance of a cancer predisposition in the family, especially among relatives who do not have cancer.

If a mutation is identified, testing of pre-symptomatic relatives can be offered to determine their risk of developing cancer. Those who have inherited the mutation can receive targeted prevention and surveillance programs. Those who have not inherited the genetic mutation do not require cancer surveillance on the basis of their family history.

Further information

Please contact the Familial Cancer Unit for information about their services and patient referral.

Familial Cancer Unit

Women's & Children's Hospital
72 King William Road, North Adelaide SA 5006
t 08 8161 6995 f 08 8161 7984
cywhs.famcancer@cywhs.sa.gov.au

A variety of pamphlets, newsletters and other resources are available free of charge from:

Cancer Council Helpline 13 11 20

8.30 am to 8.00 pm Monday to Friday

Cancer Council SA

202 Greenhill Road, Eastwood SA 5063

t 08 8291 4111 f 08 8291 4268

tcc@cancersa.org.au www.cancersa.org.au

Cancer: the significance of family history

Information for health professionals



Cancer is a genetic disease

Cancers arise as a result of the gradual accumulation of genetic errors (mutations) in several genes of a cell over time. The accumulation of these mutations can be accelerated by exposure to environmental toxins such as smoke, some chronic infections, and UV light.

This genetic mechanism is the basis for all cancer. Approximately 1 in 2 men and 1 in 3 women will be diagnosed with cancer before the age of 85.

In most people, the mutations that cause cancers are limited to somatic cells ie cells other than sperm or ova. These mutations are acquired throughout life and are neither inherited from parents, nor passed on to children.

However in 5–10% of people with cancer, one of the key mutations responsible for the cancer has been inherited from a parent. The mutation was present in the sperm or ovum from which the person developed, and was then copied into every cell of the body during development.

The presence of this cancer-predisposing mutation in every cell places the person at high risk of developing cancer at a young age. These people may have a family history of the same or related cancers. This inherited tendency to develop cancer is known as familial cancer.

What types of cancers can be familial?

It is not known why some cancers are more likely to have a familial basis than others. In general, the great majority of cancers are not familial. But some types are more or less likely to be familial.

Common cancers that may demonstrate a strong familial basis include

- breast cancer
- ovarian cancer
- colorectal cancer
- endometrial cancer.

Other common cancers such as lung cancer, leukaemia, and cervical cancer are **rarely** due to an inherited mutation in a cancer-related gene.

Some rare cancers may also demonstrate a familial tendency, including some cancers of the stomach and duodenum, retina, thyroid and brain.

There may be a familial basis for other cancers such as prostate cancer, melanoma and testicular cancer; however specific genes responsible for familial forms of these cancers have not been identified.

Risk factors for cancer

The well established risk factors for cancer include

- increasing age
- lifestyle (poor diet, physical inactivity, obesity, smoking, alcohol)
- environment (chemicals, UV exposure, infection)
- family history of cancer.

About 1 in 5 cancer patients report having a family history of cancer. This is usually due to chance, as cancer is more common with increasing age. A familial cluster of cancer can also be due to the family's shared lifestyle and exposure to similar risk factors. But a familial cluster of cancer diagnoses could also be due to the affected relatives having the same inherited mutation in a cancer-related gene.

Having a family history of cancer

It is important to identify people at increased risk of familial cancer while they are well as there may be effective risk reduction strategies and cancer surveillance methods available.

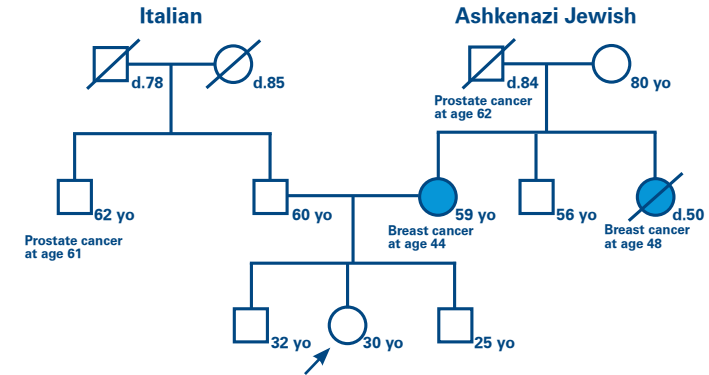
Taking a family history

The first step in identifying people at increased risk of familial cancer is to document their family history of cancer.

This history should include

- 3 generations on both sides of the family, including all first and second degree blood relatives
- all blood relatives in these generations, whether they have had cancer or not
- the current age of all relatives (if known)
- the site of primary cancer and age of onset in affected relatives
- cultural and ethnic background (some familial cancers are more common in certain ethnic groups)

A simple way of recording this information is to draw a family tree or a pedigree. The arrow indicates the person being assessed. Filled symbols identify those with breast cancer. A diagonal line indicates a deceased relative.



Pedigree of a family with familial breast cancer

Record the date on the pedigree and update it if there is a new cancer diagnosis in the family.

The pedigree provides a visual summary of the diagnoses in relatives, and makes it easier to assess the risk of familial cancer for your patient.

What to look for

Look for **any** of these features on the pedigree

- 3 or more close relatives on the same side of the family with the same (or related) cancers eg breast/ovarian, colorectal/endometrial
- any cancers occurring at an early age (less than 50 years)
- a relative with 2 primary cancers eg bilateral breast cancer, or colorectal and endometrial cancer
- an unusual cancer eg breast cancer in a male relative, medullary thyroid cancer, retinoblastoma
- the presence of multiple adenomatous polyps in a relative
- the identification of a gene mutation that confers a high risk of cancer in a relative

The presence of **any** of these features may suggest that the family has an inherited tendency to develop cancer, even if they have only been identified in one affected relative.

If the person is at increased risk of developing familial cancer, or if you want to clarify the risk, please refer them to the Familial Cancer Unit for assessment (address below), or contact the Unit for advice.