

- 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

Further information for both family members and healthcare professionals is available from the Familial Cancer Unit on 08 8161 6995 or Famcancer41300@health.sa.gov.au

Please address referrals to Dr Graeme Suthers or Dr Nicola Poplawski and send to the address below.

Familial Cancer Unit
SA Clinical Genetics Service
SA Pathology
at the Women's & Children's Hospital
72 King William Rd
North Adelaide SA 5006
t 08 8161 6995
f 08 8161 7984
e Famcancer41300@health.sa.gov.au

A variety of leaflets, 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.

Information for health professionals about familial cancers



Cancer
in the
family



Cancer Council
Helpline
13 11 20
www.cancersa.org.au

For support and information on cancer and cancer-related issues, call Cancer Council Helpline. This is a free and confidential service available Monday to Friday 8:30 am - 8:00 pm
202 Greenhill Road
Eastwood SA 5063
PO Box 929, Unley BC SA 5061
t 08 8291 4111
f 08 8291 4122
cc@cancersa.org.au
www.cancersa.org.au

Cancer Council Helpline 13 11 20
www.cancersa.org.au

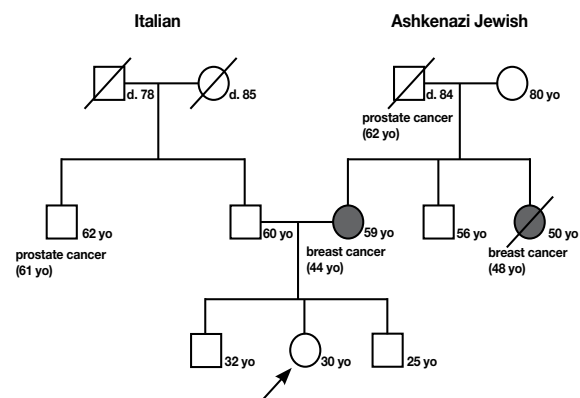
Background

1 in 2 men and 1 in 3 women will be diagnosed with cancer before the age of 85. Of those diagnosed with cancer, approximately 1 in 10 reports a family history of the same cancer in a close relative.

Significant developments in cancer genetics have occurred in the last decade. Some of the genes involved in the inherited susceptibility (or predisposition) to common cancers have been identified. As a result we have a better understanding of the cancer risk associated with a family history of the disease. Inherited mutations probably account for half of the familial clusters of cancer.

Drawing a family history

The best way to identify individuals who have inherited a genetic predisposition to develop cancer is careful evaluation of the family history. This history should include at least 3 generations of blood relatives on both sides of the family, including those with and without cancer. The simplest way of recording and interpreting this information is to draw the family as a simple diagram (pedigree). The details of each reported primary cancer site and the age at diagnosis should be documented where possible.



Pedigree of a family with familial breast cancer: the arrow indicates the person being assessed, filled symbols identify those with breast cancer, and a diagonal line indicates a deceased relative.

Identifying familial cancer

The usual clues for identifying a familial predisposition to develop cancer are

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

Management of unaffected relatives at risk of familial cancer

In a family with an inherited predisposition to develop cancer, the first- and second-degree relatives of affected family members are at increased risk of developing cancer. The management of these relatives includes

- education regarding the cancer risk
- recommendations regarding cancer prevention strategies
- implementation of cancer surveillance (where appropriate)
- referral to appropriate specialists and to a familial cancer clinic or clinical genetics service.

Familial Cancer Clinics and Genetic Counselling Services

Familial cancer clinics and genetic counselling services are available in most major cities and large regional centres in Australia. These services provide expert cancer risk assessment, counselling, genetic testing (where appropriate) and advice about cancer prevention and early detection. In addition these services often play a role in notifying at-risk relatives in the extended family of the cancer risk and the availability of counselling, testing and risk-reduction strategies.

In general it is recommended that people seeking genetic counselling have a referral from their usual medical practitioner so that the genetic management can be integrated with other aspects of their care. Many genetic services do not provide cancer diagnostic or therapeutic services and patients return to the referring doctor for continued management.

In South Australia, the Familial Cancer Unit at the Women's & Children's Hospital offers familial cancer clinics in most major public hospitals in Adelaide and in some country areas (Port Augusta and Mount Gambier).

Genetic testing in 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. This counselling involves discussing the limitations, potential benefits and possible disadvantages of testing for both the individual and other relatives. The person being tested should be prepared for the event of a positive, negative or inconclusive genetic test result.

Genetic testing can be expensive, and may take several months as 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)