



It is important that general practitioners (GPs) accurately assess a patient's personal and family history of cancer to identify those who need a referral to a family cancer clinic. Family cancer clinics will estimate an individual's cancer risk, and provide advice on risk-reduction strategies and the relevance of genetic testing.

## eviQ cancer genetics

eviQ provides current, evidence-based cancer genetics information to assist GPs in the referral process, at the point of care. Free of charge, this includes information about genetic testing and risk management, as well as information for individuals and families with a specific gene mutation, syndrome or condition.

This information is developed by the eviQ Cancer Genetics Reference Committee, which involves clinical geneticists, medical oncologists, genetic counsellors and other health professionals in accordance with the [eviQ Governance Framework](#).

The *eviQ Cancer Genetics GP Referral Guidelines*, and an outline of other cancer genetics information available on eviQ, is provided in this fact sheet.

## Purpose of the *Cancer Genetics GP Referral Guidelines*

The purpose of the guidelines is to define at-risk groups requiring referral to a family cancer clinic that offers genetic counselling and genetic testing, if appropriate.

If needed, there are two stages to genetic testing:

- **Mutation search.** This will usually involve testing a blood sample from an affected family member in the first instance. This test determines whether a gene mutation that causes the increased risk of cancer for that family can be identified.
- **Predictive test.** This is only available to family members when a mutation has already been found in a mutation search. This test determines whether or not the patient actually has the family gene mutation.

➔ Access to eviQ is free at [www.eviQ.org.au](http://www.eviQ.org.au)

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## Cancer Genetics GP Referral Guidelines

All of the people who fall into the categories below warrant a referral to a family cancer clinic for genetic counselling and risk management advice.

The categories highlighted by ● have a high probability of being offered germline testing.

The categories highlighted by ● may be offered germline testing following review by a family cancer clinic.

GENERAL PRACTITIONERS	
Individual characteristics	
Breast cancer under age 35	●
Male breast cancer	●
Multiple primary tumours (excluding lung and skin) under age 70 (e.g. breast and ovarian, fallopian tube, primary peritoneal, endometrial and colorectal cancer)	●
Colorectal cancer under age 40	●
Rare tumour* under age 45	●
Family history characteristics	
Blood relative of a known carrier of a mutation in a high-risk breast cancer predisposition gene (eg <i>BRCA1</i> , <i>BRCA2</i> )	●
Blood relative of a known carrier of a mutation in a high-risk colorectal cancer predisposition gene (e.g. <i>APC</i> , <i>MYH</i> , <i>MLH1</i> , <i>MSH2</i> , <i>MSH6</i> , <i>PMS2</i> )	●
Blood relative of a known carrier of mutation in other high-risk predisposition genes (e.g. <i>TP53</i> , <i>PTEN</i> , <i>VHL</i> , <i>SDHA</i> , <i>B</i> , <i>C</i> or <i>D</i> , and <i>NF2</i> )	●
Personal or family history of breast or ovarian cancer, and Ashkenazi Jewish ethnicity	●
Two or more first- or second-degree relatives on the same side of the family with CRC or endometrial cancer with one diagnosed under age 50	●
Two or more first- or second-degree relatives on the same side of the family with either breast cancer under age 50 and/or ovarian cancer at any age	●
Rare tumour* at any age and a close relative with similar tumour	●
Tumour pathology characteristics	
Epithelial ovarian, fallopian tube or primary peritoneal cancer ≤70 years	●
Triple negative breast cancer (TNBC) ≤40 years at diagnosis (TNBC: oestrogen, progesterone and HER2 receptor negative)	●

\**Phaeochromocytoma*, *paraganglioma*, *sarcoma*, *glioblastoma*, *choroid plexus carcinoma*, *adrenal cancer*, *retinoblastoma*

“I have had the privilege of seeing the positive influence of eviQ on clinical practice and patient outcomes over a long period of time ... It is the best example I know which truly showcases what cancer health professionals can deliver for a public good”

Founder and program director of eviQ, Professor Robyn Ward AO, Director of Cancer Services, Prince of Wales Hospital; and Board Member of the Cancer Institute NSW



## Genetic testing for heritable mutations and syndromes

For example, BRCA1, BRCA2, von Hippel-Lindau and Li-Fraumeni syndrome

### Contains information on:

- the target population
- investigations which should be considered before germline genetic testing
- factors which influence the pre-test probability of a heritable mutation
- circumstances in which testing is not indicated
- diagnostic testing
- interpretation of mutation testing results
- website resources.

## People and families with specific gene mutations or syndromes

For example, BRCA1, BRCA2, familial adenomatous polyposis and Lynch syndrome

### Contains information on:

- managing the risk
- the effect on family members
- further information and support.

**For further information on genetic testing for heritable mutations, and risk management for specific gene mutations and syndromes, please use the eviQ links below:**

### Genetic testing for heritable mutations

- [Genetic testing for heritable mutations in paraganglioma-phaeochromocytoma genes](#)
- [Genetic testing for heritable mutations in the APC gene](#)
- [Genetic testing for heritable mutations in the ATM gene](#)
- [Genetic testing for heritable mutations in the BRCA1 and BRCA2 genes](#)
- [Genetic testing for heritable mutations in the E-cadherin gene](#)
- [Genetic testing for heritable mutations in the multiple endocrine neoplasia \(MEN\) 1 gene associated with MEN type 1](#)
- [Genetic testing for heritable mutations in the PALB2 gene](#)
- [Genetic testing for heritable mutations in the RET gene associated with multiple endocrine neoplasia \(MEN\) type 2](#)
- [Genetic testing for heritable mutations in the retinoblastoma 1 \(RB1\) gene](#)
- [Genetic testing for heritable mutations in the STK11 gene](#)
- [Genetic testing for heritable mutations in the Von Hippel-Lindau \(VHL\) gene](#)
- [Germline genetic testing for hereditary mutations in the mismatch repair genes](#)
- [Germline genetic testing for Li-Fraumeni syndrome](#)

### People and families with specific gene mutations

- [Information for people and families with a faulty BRCA1 gene \(mutation\)](#)
- [Information for people and families with a faulty BRCA2 gene \(mutation\)](#)
- [Information for people and families with familial adenomatous polyposis \(FAP\)](#)
- [Information for people and families with Lynch syndrome](#)
- [Information for people and families with PTEN hamartoma syndrome](#)
- [Information for people and families with Von Hippel-Lindau \(VHL\) syndrome](#)

## Risk management for specific gene mutations or syndromes

For example, BRCA1, BRCA2, familial adenomatous polyposis and Lynch syndrome

### Contains information on:

- the target group
- exclusion criteria
- lifetime risk of cancer
- cancer risk management guidelines
- management of associated health problems and side effects
- evidence for cancer risk management guidelines
- website resources.

## Further cancer genetics referral guidelines

The following guidelines are also available for people with a personal or family history of the following cancer types:

- [Cancer genetics: Breast and ovarian cancer referral guidelines](#)
- [Cancer genetics: Colorectal cancer and polyposis referral guidelines](#)
- [Cancer genetics: Endocrine referral guidelines](#)
- [Cancer genetics: Gastric cancer referral guidelines](#)
- [Cancer genetics: Ovarian and endometrial cancer referral guidelines](#)
- [Cancer genetics: Renal cancer referral guidelines](#)

## Risk management

- [Risk management for an unaffected female ATM mutation carrier](#)
- [Risk management for an unaffected female BRCA1 mutation carrier](#)
- [Risk management for an unaffected female BRCA2 mutation carrier](#)
- [Risk management for an unaffected female PALB2 mutation carrier](#)
- [Risk management for Birt-Hogg-Dubé syndrome](#)
- [Risk management for familial adenomatous polyposis](#)
- [Risk management for Gorlin syndrome/nevoid basal cell carcinoma syndrome](#)
- [Risk management for individuals at moderately increased risk of colorectal cancer](#)
- [Risk management for individuals with a heritable CDH1 mutation](#)
- [Risk management for juvenile polyposis syndrome](#)
- [Risk management for Li-Fraumeni syndrome](#)
- [Risk management for Li-Fraumeni syndrome in children](#)
- [Risk management for Lynch syndrome](#)
- [Risk management for MUTYH \(MYH\) associated polyposis \(MAP\)](#)
- [Risk management for neurofibromatosis type 1](#)
- [Risk management for paraganglioma – phaeochromocytoma predisposition syndromes \(SDHA, SDHB and SDHC gene mutations\)](#)
- [Risk management for paraganglioma – phaeochromocytoma predisposition syndromes \(SDHD and SDHAF2 gene mutations\)](#)
- [Risk management for Peutz-Jeghers syndrome](#)
- [Risk management for PTEN hamartoma syndrome](#)
- [Risk management for unaffected male BRCA1 or BRCA 2 mutation carrier](#)
- [Risk management for Von Hippel-Lindau \(VHL\) syndrome](#)



### Additional resources:

- [Centre for Genetics Education, NSW Health](#)
- [Cancer Australia](#)
- [Hereditary Cancer Registry](#)
- [Association of Genetic Support of Australasia](#)

## Working together to lessen the impact of cancer

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