Information & Resources

Breast Cancer Network Australia

Family history and hereditary breast cancer
Fact sheet

This fact sheet is for those seeking information about family history and breast cancer.

A common perception is that a woman's risk for developing breast cancer is increased if she has even one relative affected by the disease. In reality, only 5% to 10% of breast cancer can be strongly linked to inherited factors. Most breast cancer has no association with a family link.

This fact sheet explains when family history may be important in breast cancer risk, and the options available if you have concerns about a strong family history.

Is family history important?

Yes, sometimes.

For every eight women in Australia, one will develop breast cancer before the age of 85. This makes breast cancer a common disease. It means that many women may have one or two relatives with breast cancer diagnosed at an older age (over 50 years) – through chance alone. In these cases, family history is not usually considered important for breast cancer risk.

Family history becomes more important the more relatives with breast cancer there are on the same side of the family, especially if their diagnoses occurred at an early age. In these cases, family history may indicate an increased risk for developing breast cancer. In a **very** small number of instances, it may indicate an inherited genetic risk for the disease.

What is my risk through family history?

Most women in Australia (more than 95%) have the average risk for developing breast cancer of 1 in 8. If you have concerns about your risk because of family history, a simple checklist developed by the National Breast Cancer Centre (NBCC) can help to clarify whether you have a higher risk than most.

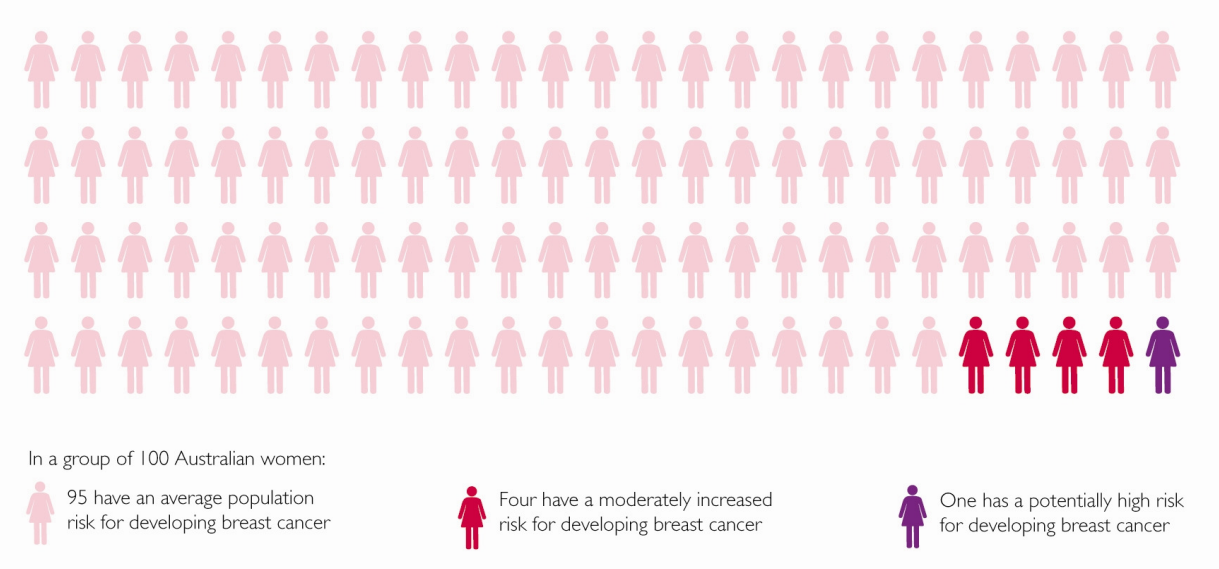
This checklist consists of a set of questions that ask about breast cancer in your mother, sisters and daughters. There are also questions about your grandmothers, aunts and nieces – on **both** your mother's and father's side of the family.

For the family history checklist go to www.nbcc.org.au/resources and use the search function.

By answering the questions, you will find you have one or other of the following risks:

- average population risk (more than 95% of females) OR
- moderately increased risk (less than 4% of females) to potentially high risk (less than 1% of females) – these terms are explained in the next section.

Compiled by Breast Cancer Network Australia
1800 500 258 www.bcna.org.au



What can I do if I find a strong family history?

Doctors have guidelines to follow to help you understand your risk and what to do next. If your answers put you in the moderately increased to potentially high risk group, you can make an appointment with your doctor. He or she should assess in greater detail your family history to determine which specific group you are in.

As a guide only (be sure to consult with your doctor) women with moderately increased risk for breast cancer tend to come from families with:

- breast cancer in two individuals on the same side of the family OR
- breast cancer in individuals before they turned 50.

Your doctor should discuss what this means for your own breast cancer risk.

It is important to note that a moderately increased risk means 75% to 90% of women in this group will **not** develop breast cancer (the lifetime risk is between 1 in 8 and 1 in 4).

Women with potentially high risk for breast cancer tend to come from families where breast and ovarian cancers are found in at least two individuals on the same side of the family, **plus** one or more of the following features:

- additional relatives with breast or ovarian cancers
- breast cancer in individuals before they turned 40
- breast cancer in both breasts in individuals
- breast and ovarian cancers in the same woman
- breast cancer in a male relative(s)
- Ashkenazi (Eastern European) Jewish ancestry.

Once again, your doctor should discuss what this means for your own breast cancer risk.

It is important to note that a potentially high risk means that 50% to 75% of women in this group will **not** develop breast cancer (the lifetime risk is between 1 in 4 and 1 in 2).

Remember, no woman – irrespective of family history – has a 0% risk of breast cancer, and no woman has a 100% risk either.

Are there specialists I can see about my risk?

If your family history means you have a moderately increased risk for developing breast cancer, your doctor should consult with a specialist genetic cancer service or Family Cancer Clinic and discuss their

advice with you. Their advice may include further assessment by specialists and a plan to monitor your health.

If your family history means you have a potentially high risk, your doctor should refer you to (or you can make direct contact with) a Family Cancer Clinic for further assessment, specialist support (information, advice and counselling) and a plan to monitor your health.

For women in both groups, a plan can include:

- Being 'breast aware' and consulting with your doctor promptly about any changes you notice about your breasts. For more on breast awareness go to www.nbcc.org.au.
- Regular mammograms and clinical breast examinations. The frequency of these tests should be tailored to your individual circumstances.

Additionally, for women with potentially high risk, a plan can include:

- Screening for ovarian cancer.
- Counselling (at Family Cancer Clinics) where you can find out more about breast cancer in families.

  If you wish, you can discuss the options of genetic testing and risk reduction as part of your plan. See the sections, 'Do I need to know if I have inherited a predisposition to breast cancer?' and 'What else can I do about reducing my risk?'

To find a Family Cancer Clinic near you telephone the Cancer Council Helpline on 13 11 20.

For Family Cancer Clinics go to www.cancer.org.au and use the search function.

What is the link between genes and breast cancer?

Genes are always involved in cancer. The cells in our bodies are constantly reproducing and cancer occurs if a cell becomes abnormal and reproduces out of control. Scientists understand that certain genes are involved because they contain important information to control cell division. If something goes wrong with such 'cancer protection genes' then cell life is affected.

Something goes wrong when genes experience small changes (or 'mutations') during cell division. Over time, these changes build up, and genes can develop 'faults'. If this happens to a cancer protection gene, then this in turn makes it difficult for an affected cell to divide and reproduce in a normal way.

Scientists point out that a certain number of different cancer protection genes need to develop faults before a cell becomes abnormal. Scientists do not know why faults develop. What they know is that they are more likely to happen the older we get and are not the result of something we have inherited.

This is why increasing age is a known risk factor for breast cancer, and why gene faults occurring over a person's lifetime affect only their own breast tissue cells and can't be passed down to their biological children. This is also why most breast cancer (90% to 95%) is not due to hereditary reasons.

What about hereditary breast cancer?

Scientists understand that a **very** small proportion of breast cancer – between 5% and 10% – is due to gene faults that are inherited. These faults are present at conception in the first cell inherited in the egg or sperm. Those inheriting the faults are known as 'gene mutation carriers'.

The two most well-known breast cancer protection genes are BRCA1 and BRCA2. The estimated frequency of BRCA1 or BRCA2 gene mutation carriers in the general population is one per 600 to 1,000 people. For those of Ashkenazi (Eastern European) Jewish descent, the estimated frequency of BRCA1 or BRCA2 gene mutation carriers is one per 50 to 100 people.

Scientists have identified a handful of other genes involved in hereditary breast cancer, such as Tp53 (gene mutation carriers in one per 10,000 people). There are likely to be unidentified genes as well, although at this stage BRCA1 and BRCA2 are the most important in terms of breast cancer risk. It is estimated that inherited faults in BRCA1 and BRCA2 cause between 1% and 5% of all breast cancer.

For more on genetic risk and breast cancer go to www.genetics.com.au (see fact sheets 44 and 45).

Could I have inherited a risk for breast cancer?

There are over 25,000 different genes! And we each have two copies of every gene in each of our cells (one from each biological parent). These include BRCA1 and BRCA2 – two cancer protection genes.

If a person inherits a faulty copy of BRCA1 or BRCA2, then their children have a 50% chance of having the same mutation that makes the gene faulty. For these family members, the risk for developing breast cancer over their lifetime is potentially very high – between 40% and 80%. There is also a higher risk of developing breast cancer at a younger age and ovarian cancer too.

Importantly, inheriting a faulty copy of BRCA1 or BRCA2 does **not** mean a mutation carrier will definitely develop breast cancer. They still have a good copy of the gene concerned. Something needs to go wrong with the good copy – and the copies of other cancer protection genes – for a breast cell to be affected, leading to breast cancer.

No woman inherits breast cancer, although a very small number inherit a genetic risk or 'predisposition' to the disease.

Do I need to know if I have inherited a predisposition to breast cancer?

Having a strong family history of breast cancer does not automatically mean an inherited genetic risk. BRCA1 and BRCA2 gene mutation carriers are rare and breast cancer is a common disease.

Should you have concerns about your genetic risk, you can meet with a counsellor at a Family Cancer Clinic to gain more information and consider your options. These include being tested for BRCA1 and BRCA2 mutations – but only if this is what you want to do.

The test involves first searching for a mutation in a living family member with breast or ovarian cancer; if a mutation is found, then testing can be offered to you and other adult relatives who may also be gene mutation carriers. Testing can indicate the presence or absence of mutations in either gene, but cannot tell whether breast cancer will or will not develop.

Knowing more about your genetic status can be a positive experience. There may be mixed feelings too, such as the worry the test results could bring to you and your family or having to deal with complex family relationships. Confidentiality, privacy and the potential to use genetic information in a negative way by third parties (such as insurers or employers) are other issues to consider.

It is crucial to discuss your options and the implications of genetic testing with your counsellor at the Family Cancer Clinic, so you can make an informed decision about what is right for you. You may also wish to have conversations with members of your family.

Some women feel it is important to undergo a test; for others, it is not so important to know. The decision to undergo a genetic test – if recommended – is entirely up to you. Pre- and post-test counselling by a specialist at the Family Cancer Clinic is crucial to support you in your decision.

BCNA has developed a position statement and background paper that covers genetic testing in more detail. Go to www.bcna.org.au and use the drop-down menu for 'Policy and advocacy'.

What else can I do about reducing my risk?

Counsellors are available at Family Cancer Clinics to discuss other options. In addition to a plan to help monitor their health, those concerned about a potentially high risk for breast cancer can discuss risk-reducing surgery.

Nothing completely eliminates the risk for breast cancer for any woman; however surgery is considered the most proven of all steps in reducing risk. Surgery includes the removal of both breasts (bilateral mastectomy). Breast reconstruction may be offered at the same time, although it is important to discuss costs involved as reconstruction can be expensive.

Surgery can also include the removal of ovaries and fallopian tubes. Around 5% to 10% of ovarian cancer is explained by inheriting a faulty copy of genes such as BRCA1 and BRCA2.

Undergoing surgery is a serious decision so it's important to discuss your options and the implications of taking these steps with your counsellor at the Family Cancer Clinic.

Remember, the decision to have surgery is entirely up to you. It is important that you have all the information you need and take the time to make the decision that is right for you.

Further information and resources

Women from families with a strong history of breast cancer, including those with hereditary breast cancer, have shared their stories with BCNA. Go to www.bcna.org.au. Family history and hereditary breast cancer is the theme of *Beacon* 38. Some of the women's stories are reproduced in this issue. Go to www.bcna.org.au.

The Kathleen Cuningham Foundation Consortium for Research into Familial Aspects of Breast Cancer (kConFab) is a national group of researchers who hope to solve questions of hereditary breast cancer. These researchers have access to breast cancer families that have agreed to participate in studies, through providing blood and tissue samples. You can find out more about the eligibility criteria and current research on their website www.kConFab.org.

In February 2007, the Minister for Health and Ageing endorsed a recommendation from the Medical Services Advisory Committee (MSAC) for interim public funding of breast magnetic resonance imaging (MRI) for high risk women. A BCNA Consumer Representative was involved in the MSAC that reviewed the literature and made the recommendation to the Minister. The technology uses a magnetic field and radio waves to produce images. It has been shown to be more effective in detecting tumours in younger women with denser breast tissue. X-ray used in mammography can often miss tumours in denser tissue.

Fact sheets produced by other organisations that you may find helpful include:

- 'Family history: An information guide for patients' by NSW Breast Cancer Institute www.bci.org.au.
- 'Breast and ovarian cancer – genetic aspects' and 'Genetics and cancer' both by Centre for Genetics Education www.genetics.com.au.
- 'Cancer: Family history and genetics' by Cancer Council Australia http://www.cancer.org.au/AboutCancer/FamilyCancers.htm
- 'Familial breast cancer', Breast Cancer Care www.breastcancercare.org.uk (click on the link to 'Information' then 'Breast cancer and breast health').

Sources used for this fact sheet

This fact sheet was prepared using a number of sources in addition to those already mentioned. These include:

- BCNA consumer representatives who provided feedback on drafts of this fact sheet.
- Associate Professor Judy Kirk, Director, Familial Cancer Service, Westmead Hospital NSW.
- BCNA members who have shared their personal experiences of family history and hereditary breast cancer.
- National Breast Cancer Centre. (2006) 'Advice about familial aspects of breast cancer and epithelial ovarian cancer: a guide for health professionals' www.nbcc.org.au.
- National Health and Medical Research Council. (1999) *Familial aspects of cancer: A guide to clinical practice*, NHMRC: Canberra.

Tell us what you think

We'd love to know what you think of our fact sheet. Please forward comments to beacon@bcna.org.au or post to 293 Camberwell Road, Camberwell, Vic, 3124.

Last updated February 2007