

## KConFaB

# Participant Information Sheet

The Kathleen Cuninghams Foundation  
National Consortium for Research on Familial Breast Cancer

www.kconfab.org

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**Local Investigator**

- *You are invited to participate in a national study concerning the familial aspects of breast cancer.*
- *If you agree to participate, personal details about you will be recorded on a database (register).*
- *The information recorded will include your name, date of birth, address, medical history of cancer, health and lifestyle.*
- *Samples of your genetic material (DNA), blood cells and tissue will be stored in a central location.*

## Background Information

Breast cancer has a tendency to run in some families. Inherited genetic factors may account for about 5% of all breast cancer. Several genes are involved in breast cancer. Women who carry a mutation (alteration) in one of these genes are at high risk of developing breast or ovarian cancer, and possibly other cancers. Men carrying these mutations are at increased risk of developing prostate cancer and sometimes, breast cancer. Both men and women may carry a mutation and pass it to their children. However, not all mutation carriers develop cancer. The reasons for this are unknown.

It is now possible to test whether a person has inherited a mutation in a cancer predisposition gene. Diagnostic testing for mutations is available through Family Cancer Clinics in most states of Australia. Diagnostic testing detects most but not all mutations in the known breast cancer predisposition genes. It is possible that some families carry mutations in other genes involved in breast cancer that are as yet undiscovered. Participation in this research study may provide the opportunity for more complete genetic testing than is generally available through the Family Cancer Clinics.

## Aims of the study

In this particular study, we want to find out:

- how often mutations in breast cancer genes occur in the Australasian population;
- the kinds of mutations that predispose Australasian families to breast and ovarian cancer;
- the risk of breast and other types of cancer
- and the age at which cancers occur
- which lifestyle and environmental factors influence the risk of developing cancer in mutation carriers and which of these factors delay or accelerate the onset of cancer.

In addition, information and samples obtained through the course of this study will be available for other approved research studies. However, the researchers will not be given information that will identify any individual or family.

## How is the study being funded?

This work is funded by the Kathleen Cuninghame Foundation for Breast Cancer Research (now known as the National Breast Cancer Foundation), and is also supported by the National Health and Medical Research Council of Australia, the National Institutes of Health of the United States of America, and the State Cancer Councils. The study is coordinated by the Kathleen Cuninghame Foundation National Consortium for Research into Familial Breast Cancer (kConFab), whose aims are to provide a resource that can be used to develop appropriate strategies for breast cancer prevention, early detection, genetic counselling and medical management of those people who carry mutations in genes that predispose to cancer.

## Who can participate in this research project?

The individuals who can best help us in this study are men and women, both with and without cancer, from families who, for example,

- have 3 or more close relatives on the same side of the family diagnosed with breast and/or ovarian cancer.  
*or*
- have 2 or more close relatives on the same side of the family diagnosed with breast or ovarian cancer including any one of the following features: breast cancer in both breasts; breast or ovarian cancer diagnosed at age 40 or younger; breast cancer and ovarian cancer in the one individual; breast cancer in a male.  
*or*
- have already had genetic testing and are known to carry a mutation in a gene involved in breast cancer.

## Plan of this study

### Questionnaires

We would like you to complete questionnaires to provide information about health, diet and lifestyle. The questionnaires will either be administered by a trained interviewer or will be mailed to you for completion and return in a reply-paid envelope. The questionnaires will take about 30 minutes to complete. Many of the questions will ask about your medical history and any history of cancer in you and your family.

We may also wish to check your medical records to confirm details about any cancer diagnosis you may have. Sometimes we verify such details against information stored in cancer registers and death records. With your permission, we would like you to tell us who your close relatives are (eg. parents, sisters, brothers, children over 18 years, grandparents), and whether or not they have had cancer. We may ask you similar questions in future. Please feel free to decline to answer any questions.

### Contacting the family

If you agree, and once you have obtained the consent of family members, we may then approach those members of your family to invite them to participate in our study. We will not reveal any of your personal details to other family members.

We ask that you try and keep us informed of any change of address or phone number. We may also ask you to nominate a friend or relative through whom we can reach you in the future if, for example, you have changed your address.

### Blood samples

We would like your permission to take 20 ml (4 teaspoons) of blood to carry out extensive testing on genes associated with breast cancer. In general, there are minimal if any side effects from this procedure; occasionally there may be some small pain or discomfort, and a small bruise may form. The blood will be stored in a central laboratory where genetic material (DNA and RNA) will be extracted, and in some cases, long-living cell lines will be established.

### Tissue samples

We would like your permission to use a small part of any tissue that has been removed in the past, or may be removed in the future and has been examined by a pathologist for the presence of cancer. Any tissue we use will be in excess of that required by the pathologist. We will use the tissue to study how cancer develops.

Sometimes we ask to obtain DNA from cells collected by a special mouthwash.

What will happen to the information and samples obtained through the study?

If you agree to participate in this study, personal details about you will be recorded on a central electronic database (register), including your name, date of birth, address and information about your medical history, health, diet and lifestyle. Samples of your genetic material (DNA), blood cells and tissue samples that have been examined for the presence of cancer (if applicable) will be stored in a central location where the database is kept.

Your privacy and other rights and interests are given the highest priority. The database will be maintained and used in accordance with very strict rules and procedures. These are consistent with the Guidelines for Genetic Registers of the National Health and Medical Research Council of the Commonwealth of Australia.

You may, if you wish, be given a copy of a document describing those rules and procedures in detail. Alternatively, you may be satisfied by the following description of their essentials.

#### Details concerning the database

One person (called the Keeper of the database) is responsible for maintaining the security of the database so that no one may have access to it except according to set rules and procedures. The database privacy policy is available at [www.kconfab.org](http://www.kconfab.org). No one will have access to your personal details, except the Keeper and the support staff, all of whom understand that they have a duty of confidentiality which must be strictly observed. In some States of the Commonwealth, statutory law covers this duty of confidentiality and there are severe penalties for breaches of the duty. The only other people who will be able to access your personal details are the interviewer and those who are providing you with genetic counselling or medical care.

A researcher who gains approval to use information or samples stored in the database may have access to your pedigree (diagram of your family tree) and the samples from you held in storage. If you have had cancer, clinical details about it and your treatment might be released. However, in most cases, researchers will not be given any information that might identify you. The information and samples are coded and in most cases, researchers will know

you only by a number. The diagram of your pedigree will have no names on it, only numbers. Only if the researcher(s) need to contact you for additional information will your information be released.

For a researcher applying to gain access to the database or to use your samples, the following requirements will be essential:

- The researcher must demonstrate that there are adequate resources, including funding, to complete the project satisfactorily. That will usually mean that there is a research grant, which means in turn that the project has been subject to strict scientific scrutiny. The project must have been examined by appropriately qualified scientists and judged to be of good scientific merit.
- The Human Research Ethics Committee of the researcher's hospital, university or institution must have approved the project.
- When these requirements are satisfied, a decision whether to grant access to the database will be made by the Executive Committee of kConFab, which is ultimately responsible for the proper maintenance and use of the database and biological samples.

If you consent to be a participant and subsequently change your mind, you may at any time withdraw from the study. In that case, if you request, all information about you will be destroyed, and you may instruct the Keeper of the database as to what is to happen to your stored material. Your instructions will be followed.

It is possible that information about the study might be published in scientific journals. However, no information that might identify you will appear in these publications.

We cannot promise that you will benefit from being a participant. We do hope and expect that the outcomes of the research made possible by the study will be of benefit to members of families like yours.

#### Results

The full results and implications of this study are unlikely to be known for several years. However,

you will receive a newsletter that will contain information about the study in a form that will not allow individuals in the study to be identified.

As part of the study, blood samples from members of your family are extensively tested for mutations. You can choose to be informed if an inherited mutation is found in your family. If you choose to be informed, kConFab will send you a letter saying that a mutation has been found in one or more members of your family. However, kConFab is not allowed to release results for individuals within the family. If, after receiving a letter from kConFab, you want to find out whether or not you have inherited the family mutation, you should contact a Family Cancer Clinic. A list of these Clinics will be enclosed with the letter, The Family Cancer Clinic, will provide genetic counselling (ie information and advice). At that stage, a second blood sample will be taken and sent to an accredited testing laboratory. KConFab may inform a Family Cancer Clinic of the general results of its genetic testing for the family. This allows the Clinic to then test individuals in your family.

If you decide at this stage not to be informed about the general test results for the family (when available), you are free to change your mind in the future. For your part, it will be necessary to keep us informed of any change of address, so that we may contact you.

Under no circumstances will anyone connected with this study reveal your information to your relatives, or any other third party without your written approval.

We encourage you to tell your General Practitioner (GP) that you have participated in the study. If you attend a Family Cancer Clinic, the Clinic may, with your permission, keep your GP informed.

As part of your medical care, your doctor may arrange for your DNA to be tested for mutations in genes that predispose to cancer. This is in addition to the research testing that kConFab will do. Sometimes the other laboratory will be first with a result. If this happens, we would like your permission for the test result to be given to kConFab for inclusion in its database. This will prevent unnecessary duplication of the testing.

**Other factors to consider before participating in this study:**

- You may find that you (and potentially other members of your family) carry a gene mutation that predisposes you to breast and ovarian cancer. The mutation may be passed on to the next generation.
- Women who carry a gene mutation are at increased risk of developing cancer but it is not currently known precisely by how much this risk is increased.
- At this time there is no treatment that can reverse the effects of mutations in genes that predispose women to breast and/or ovarian cancer.
- a level of surveillance appropriate to your age and increased risk will be discussed with you at a Family Cancer Clinic.
- We do not currently have a proven method of preventing cancer although screening of the breasts by mammography and/or other methods may detect cancers. Risk of cancer may be reduced by surgical procedures. We recommend that all participants concerned by the family history of cancer consult a Family Cancer Clinic for detailed assessment and risk management.
- Details of your family history are relevant in assessing your risk profile for certain forms of insurance. The ability to obtain health insurance is not changed by your family history or genetic test results. The Investment and Financial Services Association (IFSA) has confirmed that any existing life, trauma or disability insurance that you may have will not be affected by your participation in the study. There is a possibility that after your participation we may write to you with information that a cancer susceptibility gene mutation has been found within your family. This fact will need to be disclosed on future, new insurance applications and as such may affect assessment of your application.
- Most genetic tests cannot determine parentage. However, in some testing situations, if non-paternity or non-maternity is present, genetic testing might reveal it. This can happen when certain types of tests are done on several members of the one family. If non-paternity or non-maternity is detected, kConFab will not disclose it to you or anyone else unless required by law to do so.

- Information held by hospitals (including kConFab documents) may be subject to Freedom of Information (FOI) legislation, but personal information will not be released without consent.

### If the testing does not find a mutation in the family

- You and your relatives may still develop cancer. People who do not inherit mutations may still develop breast or ovarian cancer. The majority of those cancers are likely NOT to be due to inherited gene mutations.
- You and your family may still carry undetected gene mutations that predispose you to breast and ovarian cancer. We will do our best to search for inherited mutations. However, some mutations may escape detection or may be present in as yet undiscovered genes, and we therefore cannot guarantee to find a gene mutation should one exist in you or your family.
- You should continue to have regular surveillance as recommended by your doctor.

increased risk of cancer. A **gene test** involves analysis of one or more of those genes to determine whether a mutation is present

**Mutation:** Change (or fault) in the normal DNA code which may cause disease

**Cancer predisposition gene mutation:** Changed DNA codes which gives rise to an increased risk of certain cancers

**DNA** (*Deoxyribonucleic acid*): The chemical compound of which the genes are made **RNA** (*Ribonucleic acid*): The chemical message from the genes

**Non paternity/ Non maternity:** Is the situation in which a man/woman considered to be a person's biological father/mother is not the father/mother.

- If you consent to be a participant you will be given a copy of this form to keep.
- **CONTACT DETAILS:** If you have further questions please contact Ms Mary Anne Young at the Family Cancer Clinic at *PeterMac* on 03 9656 1064.
- You can also contact the kConFab research nurse at the *PeterMac* Family Cancer Clinic on 03 9656 1903.
- **COMPLAINTS:** Any person with concerns or complaints about the conduct of the research study can contact the Ethics co-ordinator, Mr Jeremy Kenner on 03 9656 1699 or email [jeremy.kenner@petermac.org](mailto:jeremy.kenner@petermac.org)

#### Explanation of terms used in the information sheet and consent form

**Genes associated with cancer:** Specific genes in which changes (*mutations*) have been associated with an