# kConFab

## Summer 2002 Newsletter

Published by kConFab, Peter MacCallum Cancer Institute, St Andrews Place, East Melbourne, Vic., 3002
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## Global Walk

"Walk Polly to the end of the Road" was a headline in the local newspaper during the time kConFab was holding its 2001 annual general meeting and national conference at Palm Cove, just north of Cairns.

kConFab's community representative, Mrs Gerda Evans made some enquires and located Polly Letofsky virtually outside the conference centre. Polly hails from Los Angeles and has embarked on a 4 year, 18,000 mile walk around the world to raise awareness and funds for breast cancer research.

The Australian leg of Polly's walk had taken her from Melbourne to Port Douglas. All of the money raised in Australia by Polly has been donated to the Breast Cancer Network Australia, **www.bcna.org.au** which is a national consumer organisation representing women with breast cancer.

Polly addressed the conference delegates and talked of her commitment and dedication to raise awareness and funds to support breast cancer research. The next stage of Polly's journey is to Asia.

All kConFab members wish Polly well for her breast cancer awareness campaign..

*Gerda Evans, kConFab Community Representative (left); and Polly Letofsky (right) in Palm Cove, Qld*

## Dear Readers

This is the third kConFab newsletter sent to you to thank you for your involvement with the project and to update you on our activities. This has been an important year for kConFab in many ways. Most importantly, it is the first year in which the study has been large enough to enable researchers to get stuck into some challenging and important projects. In total almost 4,500 people have contributed information and blood to kConFab, and information is complete from more than 400 families with breast cancer. The results of one of these research projects is described on page 4, (*Is BRCA3 = ATM?*), and we expect several more findings to come to fruition during the next few months.

When our research findings are published, they will be summarised on our web page **www.kconfab.org**

As a testament to the recognised excellence of kConFab as a resource for research, two large projects received funding from the National Health and Medical Research Council this year to support the kConFab Clinical Follow-up Project and the kConFab Psychosocial Study *(see Project Updates on pages 4-5)*. These successes are timely because next year the 'core' kConFab research project itself will be seeking renewal of funding in order to carry through the next five years.

kConFab itself continues to grow and now has 82 members from all over Australia and New Zealand, representing a wide range of medical and scientific specialties. There has also been some turnover at the top – Professor Joseph Sambrook resigned at the end of last year as Director of kConFab, and I became the Executive Director. This has provided some challenges because I am based in Brisbane, and the heart of kConFab is in Melbourne, but thanks to our dedicated nurses and other staff members, and Joe's continuing involvement, business is as usual.

The major contribution to kConFab of course comes from you – without the enthusiastic support of thousands of people from hundreds of families giving their time to participate, this consortium would not exist. I believe that in the next five years we will be able to make some major contributions to our understanding of breast cancer, and improve the outcome for the men and women affected.

***Dr Georgia Chenevix-Trench***
*Executive Director, kConFab*

*Below are two stories from kConFab participants who were both diagnosed with breast cancer at an early age but chose different treatment options.*

## Jane's Story

When I was diagnosed with breast cancer at 33 I couldn't believe it – breast cancer to me was something that happened to much older women. Things were happening that I had no control over and to make things worse I didn't know a thing about breast cancer. It was a time that I felt very isolated. The only other person that I knew that had had breast cancer was one of my father's sisters, who lived in WA and I didn't really know her very well.

Unfortunately during my recovery from breast cancer surgery my father's second sister was diagnosed with breast cancer and a short time after this my eldest sister was diagnosed with breast cancer (I am the youngest of four girls)! This now meant that 4 close relatives had breast cancer – what did it really mean though? This got me thinking that there may be something "there", not that I knew what "there" really meant.

In the past 7 years my life has changed dramatically and my knowledge of breast cancer and now familial breast cancer has increased enormously, in part through my association with groups like Queensland Cancer Foundation and Queensland Clinical Genetics Service.

My surgery, performed by Brisbane surgeon, Dr Furnival in 1994, went well but unfortunately for me there was node involvement. This meant six months of chemotherapy and 20 doses of radiation.
I wouldn't say 1995 was a good year for us as a family but we got through it. Luckily I didn't get sick or lose my hair. These were two things that I was sure would happen to me. That is what the general public automatically think when they hear the word chemotherapy and I was pleased to be a bit different.

In November 1999 I saw Dr Furnival again to ask him to remove my other breast which he was reluctant to do because he said that I didn't have a family history. **I had not thought to tell him about my two aunts and sister.** Once he knew of our family history he suggested that I see Dr Mike Gattas from the Queensland Clinical Genetics Service at the Royal Children's Hospital, Brisbane. Dr Gattas tested for faults in the BRCA 1 and BRCA2 genes. Surprisingly, I received a letter from Dr Gattas saying that no mutation had been found. In view of my history, Dr Gattas thinks that a susceptibility gene for breast cancer does exist in my family but at this stage it hasn't been identified. That was good enough for me. With this piece of information I went back to Dr Furnival and we agreed prophylactic mastectomy would be a reasonable decision. To have this done was not a terribly hard decision for me to make. Many of my friends thought I was very brave, but to me I would be brave if I kept my other breast in view of my family history.

When I came to my five-year anniversary I decided that I wanted to have breast reconstruction. This was one of the best things that I did, prior to this time I wasn't ready to do it, but 5 years seemed a good time for me.

A month prior to my prophylactic mastectomy I went to a familial breast cancer talk at the Queensland Cancer Foundation and meet Vivianne Geldard from the kConFab office at the Queensland Clinical Genetic Service, Brisbane. After discussion and much thought my family all decided to participate in kConFab. At this time I also arranged for kConFab to have access to my prophylactic mastectomy tissue for their research project.

I now feel I can get on and do all that I can for breast cancer support and research into this insidious disease. Hopefully the research that is being carried out will help my daughter, nieces and nephews to be free from breast cancer.

In recent years I have become a Breast Cancer Support volunteer for the Queensland Cancer Fund which is a very rewarding experience for me. I am able to visit young women who have just had their surgery. To visit other women in similar circumstances and age is very uplifting for both myself and for the patient who may think that their life is over.

I'm involved with other Breast Cancer Support women. We do talks on breast cancer to oncology nurses and radiographers.

I also helped set up a Young Women's Network (a first for Australia) through the Queensland Cancer Foundation. We decided to start up this group because young women with breast cancer face different issues to older women and these differences should be addressed.

***Queensland Cancer Foundation***
***Tel: (07) 3258 2200***

## Yvette's Story

There were not too many decisions about undergoing treatment for breast cancer at the age of 29 which I felt that I had actually contributed to, and just when I thought it better that I shouldn't think, I found myself in another overwhelming breast cancer situation - prophylactic mastectomy.

Our family has a tenacious history of breast cancer so this seemed to be the main contributing factor for the original recommendation to consider such apparently drastic surgery. Unfortunately I found myself being the target for some unwanted pressure from persuasive surgeons and concerned family before and after my lumpectomy, and although I felt that everyone had the best of intentions, eventually my stubborn nature prevailed and I was allowed some breathing space to contemplate my options.

After six months of daily hospital visits whilst undergoing chemotherapy and radiotherapy (my 6 month old son by my side), my family (living interstate) and I discussed the possibility of accessing genetic testing. Through contact with the Breast Cancer Network Australia and kConFab at a time when debates over seemingly unmanageable genetic issues were constantly being stimulated by the media, I ventured forward into this unfamiliar territory. Originally being overwhelmed about the decision that I was about to make for myself, my son and his possible future generations, Genetic Services of Western Australia placed me into their genetic counselling program. The significant amount of information and counselling that I received was excellent, and only after I remained comfortable with the knowledge of possible implications for both my family and myself was a sample taken for BRCA1 and BRCA2 genetic testing.

*Dr Georgia Chenevix-Trench, Executive Director, kConFab (left); Dr David Goldgar, International Agency for Research on Cancer, France (right)*

Whilst gaining a little thinking time while recovering from treatment, and the realisation that it could be a considerable time before my genetic test results would become available, the impending question facing me now was:

**Did my circumstances justify the need for a prophylactic mastectomy?**

I was confident that I had considered several possible scenarios.

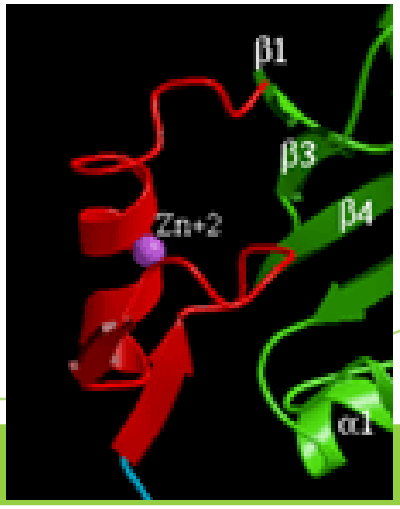
**Could my son and I handle another extension to our perilous journey so soon after being diagnosed?**

**Could the fact that I am a single parent with little support complicate recovery? Would my pending genetic test results influence my decision?**

Even though my highly aggressive tumour was detected early, evidently due to breast self-examination, attention to my possible genetic predisposition, and the availability of advanced detection techniques, my prognosis at the end of treatment was very good and almost five years on, it remains so. After further research into breast cancer, I have become aware that whatever available preventative procedure I may be able to undertake, I would never be able to achieve 100% immunity from being diagnosed with breast cancer again in my lifetime.

Currently, my genetic testing results remain inconclusive and I am content with my chances in life. Should I endure another battle of breast cancer in the future, I will contemplate the possibility of prophylactic mastectomy once again, considerably more confident in my ability to achieve wisdom and enlightenment. In what, so far, was one of the most powerful times of my life, I am convinced that I was true to myself and this significant decision was made by that of an informed, empowered breast cancer consumer.

***W.A. State Representative***
***Breast Cancer Network Australia***
***Tel: (08) 9305 2818***



## kConFab finds a new familial breast cancer gene: Is BRCA3 = ATM?

Although it is likely that most of the families enrolled in kConFab carry a mutation (fault) in a gene that predisposes them to breast cancer, hospital laboratories around the country who search for mutations in the genes BRCA1 and BRCA2 have only been able to find mutations in about a third of families enrolled in kConFab. In the *kConFab 2000 Newsletter*, Dr Southey described the search for BRCA3 using material from the families with breast cancer in which no mutations have been found in BRCA1 or BRCA2. The article mentioned a possibility of a new gene which had been reported by Scandinavian researchers. A year later it is clear that this possible new 'BRCA' gene does not seem to be involved in many, if any, families around the world.

However, in the meantime kConFab researchers have made a surprising discovery. Dr Georgia Chenevix-Trench and her colleagues at the Queensland Institute of Medical Research, have been looking at a gene called 'ATM', which stands for 'ataxia telangiectasia mutated'. This gene has been a suspect for 'BRCA3' for about 20 years (long before BRCA1 and BRCA2 were identified) because women who inherit one mutation in the ATM gene seem to be slightly more likely to develop breast cancer. Despite this rather vague association, to date no one has looked properly for mutations in ATM in families with breast cancer (probably because the previous research did not indicate that the mutations could be severe enough to result in families with multiple cases of breast cancer). Dr Chenevix-Trench started doing this in late 2000, concentrating on just two mutations in the ATM gene which she thought might be particularly important in increasing a woman's risk of breast cancer. She has focused her search on 150 families enrolled in kConFab that do not have any cases of ovarian cancer or male breast cancer within them, because it is particularly likely that these 150 family histories are due to 'BRCA3' and not to undiscovered mutations in BRCA1 or BRCA2.

So far she has found that 5/150 of these families, in which no mutation could be found in BRCA1 or BRCA2, carry one of these mutations in the ATM gene. This means, that for these families at least, there is finally an explanation for the many cases of breast cancer that have occurred within them. This information is currently being relayed to the individuals in those families who have indicated that they wanted to know of any pertinent, new information that kConFab research found. As with mutation testing in BRCA1 and BRCA2, these results will allow the individuals in these families to make much more informed decisions about the management of their cancer risks. This research will be published in February 2002. Full details will be on the kConFab web site.

If only two specific mutations in the ATM gene are responsible for about 3% of breast cancer families, it is highly likely that other mutations in the same gene might be responsible for others. Because the ATM gene is so huge (much larger than BRCA1 or BRCA2), Dr Chenevix-Trench has teamed up with Professor Michael Stratton at the Institute for Cancer Research, UK, who has the technical capacity to search for mutations in the whole gene in all the other kConFab families in which no mutation in BRCA1 or BRCA2 has yet been found.

This research has been very encouraging to the many kConFab researchers. We are doing one of the things we set out to do, namely finding mutations in genes that would never have been examined by a hospital laboratory.

We are very grateful to the many families in kConFab who have made this work possible, and to the research nurses and database support staff who have worked so hard. We hope that this research will provide answers for more kConFab families in the future, and to other families with breast cancer from around the world.

## kConFab Psychosocial Study

The kConFab Psychosocial Study, which is exploring the impact of stress and coping on the development of breast cancer, is underway. We now have a team of interviewers (Mariette and Jacqueline) and have successfully completed 20 phone interviews with new kConFab participants about stressful situations and events in their lives over the previous three years. The women participating have reported that the interview has provided an opportunity for them to tell their story and has allowed them space and time to reflect on their experiences over the past few years. Some were surprised at how much had happened, and in retrospect, were amazed at how well they had coped. About 500 kConFab participants have also filled in questionnaires, asking about emotional responses and coping. The research team is about to contact people who joined kConFab three or more years ago, about participating in the study.

Should you have any questions about the study please do not hesitate to contact me on the telephone number listed below.

***Ms Melanie Price, Study Coordinator***
***Tel: (02) 9926 7726***
***Email: melaniep@med.usyd.edu.au***

## Altered hormone action in breast tissue from women with hereditary breast cancer

Women who carry mutations in one copy of a breast cancer susceptibility gene, such as BRCA1 or BRCA2, are known to be at increased risk of breast cancer, often at an early age. The genetic alteration in the normal breast of these women may represent the first "hit" in the cancer-forming pathway and we believe that this may be accompanied by, or lead to, further changes in their normal breast cells.

Some women taking part in kConFab, who have been found to have a BRCA1/2 mutation, or who are thought to carry such a mutation, elect to have prophylactic (preventative) mastectomy and have donated some of their normal breast tissue (removed at the time of surgery) for research. We have embarked on a study to compare this normal breast tissue with normal breast tissue from women with no known family history of breast cancer. Expression of a range of biological markers that influence the behaviour of tissues were similar in both these groups of women, with the exception of a significant decrease in the expression of progesterone receptor (PR), which we observed only in the breast tissue of women from the BRCA1/2 group.

PR is a protein that is closely involved with the hormone response of tissues such as the breast. Normal breast function is dependent on the coordinated activity of hormones (oestrogen and progesterone) that are finely balanced, but change at puberty, during the monthly cycles and at menopause. Our results suggest that there may be changes to this coordinated hormone activity in the normal breasts of women who have a genetic tendency to developing breast cancer. Lack of PR in the normal breast tissue may result in heightened sensitivity to oestrogen, so this work may provide one clue as to why women who carry BRCA1/2 mutations are at increased risk of breast cancer. We now need to find out whether the altered progesterone activity we have observed is contributing to increased breast cancer risk.

The importance of the efforts of kConFab and the women participating in the kConFab study who have donated breast tissue for our study cannot be overstated and we are very grateful for their willingness to donate tissue. kConFab participants are contributing in a very real way to improving our understanding of breast cancer in high-risk women, but this important work may also eventually contribute to our understanding of breast cancer development in the broader population of women at risk of this disease.

***Dr Patricia Mote, Dr Judy Kirk, Dr Jenny Leary, Dr Kerstin Sandelin, Dr Christine Clarke,*** *Westmead Institute for Cancer Research, University of Sydney and Familial Cancer Service, Westmead Hospital, Westmead, NSW, Australia and Department of Surgical Sciences, Karolinska Hospital, Stockholm, Sweden*

## kConFab Clinical Follow-up Project

The Clinical Follow-up Project is underway with funding from the Australian National Health and Medical Research Council. We plan to mail follow-up questionnaires to all kConFab participants starting from December 2001.

The questionnaire will be sent out to each person approximately 3 years from the date of initial interview. This means that they and members of their family may receive the questionnaire at different times.

The questionnaire will collect information about health and lifestyle over the last 3 years. It asks about the sorts of health checks there may have been, about environmental factors such as exercise, alcohol and cigarette smoking, and also updates us with regard to any recent diagnoses of cancer in the family.

Don't worry if you need some assistance completing the questionnaire. It is only a phone call away. Our kConFab research assistants, Prue and Ailsa, are available on the **toll free help-line** to answer any questions. The help-line telephone number is printed on the front of the questionnaire.

We will also be sending you information about the Psychosocial Study, also featured *(see page 4)*.

***It is most important that we don't lose track of you, so if you are changing address in the near future please let kConFab know.***

We are very excited about this next phase of the kConFab study. By collecting information every 3 years (subject to funding) we hope to be able to determine which lifestyle factors influence the risk of cancer. Ultimately this should help us to better understand how to prevent cancer, or at least how to detect it early, which in turn should make it easier for you and your doctors to make decisions about your lifestyle and medical issues, and help you to live better and longer.

***Ms Prue Weideman***
*Clinical Follow-up Coordinator*

*Associate Professor Christine Clarke, Westmead Institute for Cancer Research, Sydney (left); Jenny Leary, Familial Cancer Laboratories, Westmead Hospital, Sydney (right)*

***How does a family history affect my risk?***

While the main risk factors for breast cancer are being female and getting older, family history is also a well-recognized risk factor. Because breast cancer is a common disease, familial clusters of two or more cases may occur by chance. It has been estimated that a woman who has an affected first-degree relative (mother, sister or daughter) has an approximately two-fold increased risk of breast cancer over her lifetime. The National Breast Cancer Centre (NBCC) has developed guidelines for health professionals to estimate the risk for women who are concerned about their family history. For example, a woman who has a mother affected by breast cancer diagnosed over the age of 50 would be considered to be at or only slightly above the population risk for breast cancer. More than 95% of the female population fall into that category. At the other extreme, some women are at potentially high risk of breast cancer because they have three or more affected female relatives on the same side of the family. In addition, there may be other high risk features such as breast cancer diagnosed before the age of 40. Sometimes, in those "potentially high risk" families, there are also women who develop ovarian cancer. These are the rare families who may carry a genetic (inherited) susceptibility to breast/ovarian cancer.

***What sort of a family history indicates a possible high risk?***

Women at potentially high risk of breast cancer tend to have multiple affected family members in different generations that have been diagnosed with either breast or ovarian cancer, often at an early age. A strong genetic susceptibility to breast cancer is very rare, accounting for less than 5% of all breast cancer.

***Is a genetic susceptibility more common in some ethnic groups?***

Research has demonstrated that a genetic susceptibility to breast and ovarian cancer seems to be more common in women of Ashkenazi Jewish descent. It has been found that for Ashkenazi men and women, approximately 1 in 50 or 1 in 100 individuals carry a gene mutation (fault) that puts them at higher risk of cancer.

***Now I have had breast cancer, what should I tell my daughter/s/family/relatives?***

At the time of diagnosis, although your relatives would be most concerned about you, some of them may also be concerned about how your diagnosis may affect their risk. You may be worried about the risk for your sister(s) or daughter(s). They would be best advised to see their general practitioner who can consult the family history guide provided by the NBCC and determine whether the family history is of concern. If the family history indicates that some members of the family may be at potentially high risk, then referral to a specialist Familial Cancer Service might be discussed.

***In my family there is a strong family history (4 close relatives with breast cancer). We would like to think about genetic testing. How does it work?***

When the family history is strong enough to indicate a genetic susceptibility, genetic testing may be helpful. The potential benefits, harms and limitations of current genetic testing need to be discussed in detail at this time. Thought needs to be given to the possible medical, psychological and social consequences of testing for those already affected by cancer and those who may be at risk. Then, if it is decided to go ahead with genetic testing, the first step is to take blood from one of the living, affected family members. This is usually a family member who has been diagnosed with either breast or ovarian cancer. A blood sample is sent to the laboratory and extensive testing is done to search for a fault in one of the two main genes that are known to be involved in breast cancer susceptibility. These genes are called BRCA1 (short for breast cancer 1) and BRCA2 (short for breast cancer 2). This testing may take 6 to 12 months or even longer to complete. If a gene mutation is identified in a research study, (such as kConFab), a second blood sample will be need to be taken and the research results verified in a NATA accredited laboratory.

***My family has a strong family history and so we had testing for BRCA1 and BRCA2. They have tested my mother (who had breast cancer at the age of 40) and they found no gene mutation. What does this mean?***

Unfortunately, at this stage, it is not possible to find a breast cancer associated gene fault in some families. This is either because the family history has occurred by chance or may be because the current methods of testing don't find all of the mutations (faults) in BRCA1 or BRCA2. In addition, it is quite possible that other breast cancer susceptibility genes will be found over the next few years. Testing on those "yet to be discovered" genes may be possible in the future as knowledge and testing systems improve. There is still a lot of research to be done in this field *(see page 4: Is BRCA3 = ATM?).*

***In my family my sister (who has breast cancer) has been told that she has a BRCA1 gene mutation. What will it mean if I test negative for that mutation?***

Even if a mutation has been found in your family, if you have a blood test that shows that you do not carry that same mutation, then you are not considered to be at increased risk for breast or ovarian cancer. In addition, it is important for you to know that if you do not carry the family BRCA1 mutation, then you cannot pass it on to your children. For many women this is extremely reassuring.

***What will it mean if I test positive for that BRCA1 mutation?***

If the family mutation is known and you are found to carry the same mutation, then it will mean that you are at much higher risk of breast and ovarian cancer than other women in the community. Women who have inherited a mutation in BRCA1 or BRCA2 are at a 10 to 20-fold increased risk of breast cancer. It is very difficult to be precise about the exact risk of breast cancer as this may vary according to the gene and may also depend on the specific gene fault that has been found in the family. In addition, there will almost certainly be other genetic or environmental factors that influence or impact on that risk. After a woman tests positive for such a mutation, she must consider, along with her doctors, the available management options. These include careful breast cancer screening and possible participation in a breast cancer prevention trial. Some women, after careful counselling, consider prophylactic (preventative) mastectomy as their best option. Prophylactic mastectomy has now been shown to reduce the risk of breast cancer in women at potentially high risk. Surgery to remove the ovaries (prophylactic oophorectomy) may also be considered. Individual counselling is very important at this stage as surgery is not the best option for every woman, and the benefits and risks involved for each option need to be fully discussed.

***What if my brother also wants a test for the BRCA1 mutation that runs in our family?***

Once a family mutation has been identified, all at-risk adult family members can be tested. Men who test positive for a BRCA1 gene fault seem to be at a slightly higher risk of prostate cancer later in life. As research unfolds, it may be possible that there are other cancer risks, though not particularly high. Men who test positive may also pass the mutation on to their children. For each child (male or female) there is a 50-50 chance that the mutation will be passed on. A man or woman who tests negative for a known BRCA1 mutation cannot pass that mutation on to their children.

***What happens at a Familial Cancer Service?***

If you are referred to a Familial Cancer Service, you will first see a Genetic Counsellor or Nurse who will take an accurate and extended family history, noting all affected and unaffected relatives, the age of onset and type of cancers, on both sides of the family. Sometimes the counsellor will need permission from the family to verify the history through doctors or medical records. It is on this basis that an assessment is made as to whether genetic susceptibility is likely. If genetic testing may be of benefit to the family, then genetic testing will be fully discussed. It is important for the family to understand the limitations of genetic testing as well as the possible benefits. They need to consider the possible medical and psychological implications of a positive or negative (or uninformative) genetic test. Generally, the family is given time to discuss this before a decision is made to proceed to genetic testing. When blood is taken to search for a mutation in one of these genes, there may be a long wait before any results are issued. If a mutation is identified, then other family members can be tested. This is a relatively quick and simple test. However, for each family member, there will be different issues to consider in deciding whether to have a test. This is why counselling is so important both before and after a genetic test.

***What are we doing about research in this field?***

Australia has a very good program of research in the area of familial breast cancer. Professor John Hopper from the University of Melbourne pioneered some of this work. He conducted a study of women with breast cancer to determine the importance of these gene faults in Australian women with early onset breast cancer. Following this study, a consortium of researchers was established which is now known as kConFab, short for "The Kathleen Cuningham Consortium for Research into Familial Breast Cancer". kConFab brings together geneticists, oncologists, surgeons, genetic counsellors, psychosocial researchers, pathologists, epidemiologists, nurses and other health professionals from around Australia. The aim of kConFab is to develop a resource of biological samples and other information that can be used for future breast cancer research. A number of important studies are already underway. Many Australian breast cancer families are therefore already assisting the work of kConFab through their participation. More information can be found by visiting the kConFab web page at **www.kconfab.org**

***Who can I speak to further about this?***

If you are concerned about your family history, first consult your general practitioner. You may wish to discuss referral to a Familial Cancer Service *(see page 10 overleaf)*.

***Associate Professor Judy Kirk***
***Senior Staff Specialist***
***Westmead Hospital, Sydney***

## Fifth Australian Conference on Familial and Genetic Aspects of Cancer

5 - 6th July 2002, South Australia.
For more details contact:
Dr Teresa Fisher
Email: *teresaf@nbcc.org.au*

## The NHMRC Familial Cancer: Clinical Practice Guidelines

Now available from the Australian Cancer Network.
Tel: (02) 9380 9177 for details.

## Planned kConFab Projects

A number of new research projects are still in the planning stages.

One such project will be aimed at trying to understand the importance of 'variants' (different to a normal gene) in the BRCA1 and BRCA2 genes which have been identified in kConFab families, but for which there is no clear evidence as to whether they are mutations (faults) that are related to the cancers in the family, or whether they are simply rare but benign (not detrimental to your health) variants in the gene. Currently there are about 20 families enrolled in kConFab who carry variants of this nature, and it is hoped that this research will allow the clinicians and families to make more informed decisions about the importance of these variants, and whether they can be used for guiding management decisions.

## Upcoming kConFab and international study for women at increased risk for ovarian cancer

It is hoped that a new study will be available in 2002 for kConFab participants who have an increased risk of ovarian cancer and who live in Melbourne or Sydney.

The study will compare the outcomes for women who choose preventive ovarian removal versus those who choose regular screening tests for ovarian cancer. The study will be undertaken in conjunction with researchers in the United States, and aims to include several thousand women from around the world. It is anticipated that the results of this local study will eventually assist all kConFab families in their decisions about screening and prevention of ovarian cancer.

Keep your eye on the kConFab website **www.kconfab.org** over the next few months for further details.

*Professor Michael Owen, University of Wales, College of Medicine, Cardiff, UK, Guest Speaker at kConFab 2001 AGM (left); Associate Professor Michael McKay, Radiation Oncologist/Research Scientist, Peter MacCallum Cancer Institute. Melbourne (right)*

*Dr Bettina Meiser, Prince of Wales Hospital, Sydney*

*Associate Professor Judy Kirk, Senior Staff Specialist, Westmead Hospital, Sydney*

*In order to keep kConFab running smoothly, we would greatly appreciate if you would consider the following.*

- ***Please remember that fresh surgical specimens are extremely valuable for research.*** The reason for this is that it is possible to look at how genes are actually working through the analysis of freshly frozen surgical material, in a way that is not possible using archival material that has been stored away by pathologists. There are several research projects that currently use this material, for example to find out which genes are active in the normal breast from women carrying BRCA1 or BRCA2 mutations *(see page 5)*. This will allow us to develop a better understanding of how the tumours develop, and perhaps eventually to find ways of changing the tumour development. In addition, we want to find out whether tumours from women with BRCA1 or BRCA2 mutations are different from tumours from women with as yet unidentified mutations in other genes such as ATM *(see page 4)*. This may allow us to find additional, new breast cancer genes by grouping together families with tumours that share particular patterns of gene expression.

- ***Please ring the cancer specialist at your Family Cancer Clinic to inform them of any new cases of cancer in your family.***

- ***Please ring your local kConFab Research Nurse if you, or any member of your family, is having breast or ovarian surgery.***

- ***Please tell your research nurse if you change your address***

## Cancer news on the net and other related newsgroups:

**Facing our risk of cancer**
http://facingourrisk.org

**kConFab**
http://www.kconfab.org

**National Breast Cancer Centre**
http://www.nbcc.org.au

**National Breast Cancer Foundation**
http://www.nbcf.org.au

**Breast Cancer Network Australia**
http://www.bcna.org.au

*Professor Graham Mann, Westmead Institute for Cancer Research, Westmead Hospital, Sydney (left); Dr Eric Haan, Clinical Geneticist, Women's and Children's Hospital, Adelaide (right)*

*Dr Hilmi Ozcelik, Scientist, Mount Sinai Hospital, Toronto, Canada (left); Andrea Tesoriero, Research Scientist, University of Melbourne (right)*

# Collaborating Family Cancer Centres

**Victoria**
Familial Cancer Centre
Peter MacCallum Cancer Institute
St Andrews Place
East Melbourne, 3002
Contact: Dr Sue Anne MacLachlan, Ms Mary-Anne Young
Tel: (03) 9656 1064
kConFab Research Nurse: Ms Janine Furmedge
Tel: (03) 9656 1903

Royal Melbourne Hospital
Familial Cancer Centre
Parkville, 3050
Contact: Dr Geoffrey Lindeman, Dr Clara Gaff
Tel: (03) 9342 7151
kConFab Research Nurse: Ms Marianne Griffen
Tel: (03) 9342 4257

Victorian Clinical Genetics Service
The Murdoch Institute
Royal Children's Hospital
Parkville, 3050
Contact: Dr Mac Gardner
Tel: (03) 8341 6293
kConFab Research Nurse: Ms Janine Furmedge
Tel: (03) 9656 1903

Genetic Health Services Victoria
Monash Medical Centre
Clayton, 3168
Contact: Ms Tarli Hall
Tel: (03) 9594 2026
kConFab Research Nurse: Ms Janine Furmedge
Tel: (03) 9656 1903

**New South Wales**
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Patients of Concord Hospital can ring Westmead for appointments.

Prince of Wales Hospital
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Hereditary Cancer Clinic
Kogarah, 2217
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The John Hunter Hospital
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**South Australia**
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**Tasmania**
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The Launceston General Hospital
The North West Regional Hospital, Burnie
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