



# Gene Trek

Familial Cancer Unit Newsletter



October 2006



## From the editor....

Dear Readers,

Welcome to the second issue of the GeneTrek for 2006! This is my first issue of GeneTrek as editor having recently replaced Clara Tait as the Cancer Genetics Education Officer at The Cancer Council South Australia. One of my important jobs is to assist the clients of the Familial Cancer Unit to be informed and updated on issues surrounding familial cancer. Education plays a critical role in the prevention of all cancers. In the GeneTrek we endeavour to bring you relevant and up-to-date information on subjects that are important to you. In this "new look" issue our feature article is on genetic discrimination and familial cancer, we have our regular update from the Familial Cancer Unit and a healthy and delicious cake recipe. I hope you enjoy reading this issue, and please feel free to contact me with suggestions for articles or ideas on what you want to be educated about. I look forward to hearing from you!

Kind regards

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## Genetic discrimination and familial cancer:

A report from the Genetic Discrimination Project

This report presents research findings from the Genetic Discrimination Project that may be of interest to you. More details about the GDP can be found at <[www.gdproject.org](http://www.gdproject.org)>.

500 people were surveyed during 1998-2003 to determine the type and level of discrimination they had experienced because of their genetic test result. The study was limited to unaffected people who sought genetic testing to clarify their own risk of developing a familial disorder. Of these, about one third were people at risk of familial cancer.

1 in 10 people (10%) reported negative treatment. Most problems concerned death cover, trauma and income protection insurance. Some people reported that they

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had limited or no access to life insurance because of their family history. Others gained approval for insurance but with special conditions that seemed unreasonable (to them). People with abnormal genetic test results had difficulties changing their pre-test insurance products, the amount of pre-test cover, or their insurance provider. About 1 in 7 of all people did not bother or were advised not to bother applying for life insurance because it was assumed their applications would be rejected. Some success was gained when an insurance decision was questioned or reviewed (often with support from a medical specialist presenting their real risk or documenting their screening and surveillance practices). Others were unable to reverse decisions including denial of insurance and exclusion of all cancers from claims for trauma insurance.

People also experienced negative treatment within the family and in healthcare settings. Some described negative attitudes or treatment in family settings after deciding to be tested or after receiving a test result (either abnormal or normal). Others reported negative attitudes of health professionals, receiving unsuitable clinical advice or having genetic information shared without consent. Some felt that they were under pressure to undertake testing in the genetics clinic.

Some conclusions drawn from the study:

- Most of the people surveyed did not receive negative treatment due to their family history or mutation status for a familial disorder.
- People who want life insurance products should apply for them rather than assuming that their applications will fail. (Although life insurers in Australia can legally use family history or genetic test results for assessing applications, they must also, according to the law, be able to actuarially justify their decisions).
- An unfair or unreasonable decision should be discussed with an appropriate medical specialist as well as the insurance company (especially for familial cancers) because the genetics of familial cancers and of each family history is individual and can be complex.
- People did report negative treatment by clinical genetics service providers and other health professionals.

The Human Genetics Advisory Committee has been established by the Federal Government to examine and monitor the ongoing development of genetic technologies in Australia and their implications for people with risks or family histories of inherited disorders like familial cancers. The committee's website is <[www.nhmrc.gov.au/about/committees/hgac/](http://www.nhmrc.gov.au/about/committees/hgac/)>.

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Editors note: If you would like the full length version of this article, please contact Carolyn Harrington.

## Counsellors' page



Debbie

Jacquie

Vanessa

Sally

Welcome to Counsellor's page and Ask a counsellor. Genetic counsellors Debbie, Jacquie, Vanessa and Sally bring you up to date with the latest news from the Familial Cancer Unit. If there are any queries about the information in these sections please contact them on (08) 8161 6995.

### BRCA support groups

Two years ago a needs survey was undertaken with the Familial Cancer Unit clients. The feedback indicated a need for further information and support for women who (1) tested positive for BRCA mutations and (2) people with FAP and those caring for children with FAP. In response to this need, support groups run by two genetic counsellors were piloted in the first half of 2006. The topics discussed in the support sessions are presented below.

A group for BRCA positive, unaffected women was attended by 13 women aged from 25 to 61 years (most in their 30's and 40's).

There were 4 participants in the FAP support group, with the age range of 28–50 years. Three of the participants were affected and one was caring for a child with FAP. Whilst all the participants in the FAP group were women, their partners were invited, and did attend one session where the focus was on communication with partners and family.

#### BRCA topics

- genetics revisited
- living with risk and uncertainty
- coping strategies
- family communication
- prophylactic surgery
- breast reconstruction options
- future directions in breast cancer prevention, surveillance and treatment

#### FAP topics

- genetics and APC mutations revisited
- living with FAP and caring for yourself
- talking to family members including children with FAP

Positive feedback from all participants in both groups has indicated the value of meeting with other similarly affected individuals. We have recently commenced a second group for women with a BRCA mutation. The BRCA and FAP

support groups will continue in 2007, and support groups for individuals with other conditions are planned.

## Conference highlights – August 2006

Sally and Vanessa attended the 11th International Congress of Human Genetics in Brisbane. Two international speakers presented their ideas and experience working in the field of genetic counselling at an Australasian Genetic Counsellors workshop.

Debbie and Jacquie attended the Familial Cancer Research and Practice conference at Couran Cove in Queensland. Jacquie presented the SA experience of pilot support groups for carriers of BRCA and APC gene mutations.

## High risk cancer clinics

Two clinics for women at high genetic risk of breast and ovarian cancer have begun at the Queen Elizabeth Hospital and the Royal Adelaide Hospital. These clinics will enable women to be reviewed in one place by breast and gynae-oncology specialists and to have access to a genetic counsellor at the same time. If you would like more information about these clinics contact Karen Redman (QEH) on 8222 6000 or Joya McCormack (RAH) on 8222 5917.



## Ask a counsellor

Is there an issue relating to familial cancer that you would like discussed? Write in to Ask a counsellor.

### A client writes....

*"I was tested a while ago for a breast cancer gene that had been found in our family. Although I thought I had really considered the impact of finding out I had the mutation, I am surprised at how much I am preoccupied with a sense of living with uncertainty and my risk of developing cancer."*

### Sally's response:

Being identified as a carrier of a breast cancer gene represents a disruption to the usual pattern of life, causing feelings of uncertainty and of being at risk. For most people, a period of psychological adjustment is required.

Normal responses may include:

- not thinking about your carrier status until the next surveillance
- thinking about the genetic error often, with sometimes an overwhelming and intrusive level of fear
- making the experience positive by effecting some beneficial lifestyle changes

The sense of uncertainty may be compounded by decisions about prophylactic surgery, when and what to tell other family members, and concern about passing on the genetic error to children. When aspects of life are not going particularly well (e.g. relationship difficulties, employment hassles, financial struggles), we can feel a general sense of being distressed and overwhelmed. Living with uncertainty affects people in different ways and can be related to your personality and coping style.

What you **can** do about living with uncertainty and the sense of being 'at risk'

- Challenge negative and unhelpful thoughts (gene mutation does not equal cancer). Recall the reasons for undertaking the test.
- Take one day at a time. Focus on what you can achieve today rather than possible challenges ahead.
- Plan ways to enhance your feelings of control. Plans may include setting surveillance dates, when and if you wish to discuss preventative surgical options, or deciding not to make a decision for 5 years.
- Ask questions and discuss options at medical appointments, you will feel more in control and less 'at risk'. Ask friends/family to come to difficult appointments.
- Seek professional help or an outside person if you are worried about overburdening those close to you. Reach out for support to others in similar circumstances (support groups).
- Recognise feelings/triggers of anxiety or stress and develop strategies to manage them.
- Find a positive meaning in the genetic testing; use it as an opportunity for change, for identifying the values and priorities in your life, and for deciding what gives meaning in your life, and why.
- Maintain self-esteem, create time to pursue relaxation, pleasurable activities or hobbies.
- Most importantly reduce expectations on yourself - give yourself permission to have an 'off' day!

It can take time to adjust to living with an increased risk of cancer and the important people in your life can also be affected. We are aware that issues can arise weeks, months or even years later and we are very happy to hear from you. If there are any issues that you would like to discuss in more detail, for example surveillance recommendations, prophylactic surgery or exploring your feelings, please feel free to contact us on 8161 6995.

# Healthy eating

## Reducing your cancer risk

There are lifestyle strategies that you can put in place that can help reduce cancer risk. Reducing alcohol consumption, stopping cigarette smoking, healthy eating and undertaking regular exercise can all have a positive impact on cancer risk reduction. The Cancer Council South Australia recommends people:



- Eat 5 serves of vegetables every day
- Eat 2 serves of fruit every day
- Eat no more than 3-4 serves of red meat a week and limit consumption of processed meat
- Be active every day
- Maintain a healthy body weight
- Limit or avoid alcohol.

For more information contact **The Cancer Council Helpline 13 11 20** and ask for a copy of the new LiveSmart® lifestyle guide.

## Rosalie's mango fruit cake

This is a moist and healthy fruitcake, with no added sugar, low on fat but high on flavour. Enjoy!

### Ingredients:

1 x 425g tin of mango slices in natural juice	2 eggs, lightly beaten
500g mixed fruit	pinch of salt
½ cup water	½ tspn mixed spice, optional
1 ½ tsps bicarbonate soda	¼ cup chopped nuts, optional
1 ½ cups self-raising flour, sifted	

### Method:

Spray a square or round 20 cm tin with cooking oil spray, and line with baking paper. Preheat oven to 160°C (conventional oven).

Combine mixed fruit, mango slices with juice, and water in a saucepan. Bring to the boil, and then simmer for 1–2 minutes. Remove from heat and cool to around room temperature. Stir in the sifted flour, bicarbonate soda, salt, eggs, spice and nuts (if using). Pour mixture into the prepared tin and cook for about 1 ½ hours (depending on oven). After about ¾ hour cover the cake loosely with alfoil to prevent top from burning. When cake is cooked, leave covered with foil and cool completely in the tin. Keeps well in an airtight container.

Editor's note: mango slices can be substituted with a can of crushed pineapple in unsweetened juice.

# Genetic testing in research studies

Many of the clients of the Familial Cancer Unit have agreed to assist in research studies of familial cancer. To date, over 3000 clients and relatives have assisted in this way (thank you!). The research studies are usually part of Australia-wide projects and involve laboratories in other states or countries. If a research study identifies genetic information that may be relevant to specific families, they write to the people who have been involved to advise them of this. However in many cases, clients and relatives have already been informed by the Familial Cancer Unit.

With both the Familial Cancer Unit and the research study writing letters informing families about important genetic information, there is the potential to cause confusion. A problem occurs if only one group sends letters - there is a risk that some people will miss out on hearing important news that may be lifesaving.

So if you receive a letter from a research study that tells you something you already know, there is nothing further for you to do. If you are uncertain about the significance of a letter you get from a research study (or from the Unit), please call us on 08 8161 6995 or [cywhs.famcancer@cywhs.sa.gov.au](mailto:cywhs.famcancer@cywhs.sa.gov.au). We would be pleased to clarify the situation.

***Try to remember...  
Uncertainty may be a part of your life but it does not need to affect your quality of life. If you live with uncertainty about the future it can motivate you to make the most of every day!***

## Your place

This section is dedicated to personal stories from men and women who have, or are at risk of inherited cancers. If you have a story you would like to share, please contact one of the genetic counsellors at the Familial Cancer Unit.

*My name is Adrian. I was 29 years old and entering a busy time at work and home, and we were expecting our first child. I was suffering from a few headaches and was always feeling worn out. I had visited my doctor, and my headaches were thought to be due to stresses at work. As the headaches continued over the next 12 months I was sent for a CAT scan. The test results revealed a shadow on my brain, and I was sent on the same day to see a Specialist at The Royal Adelaide Hospital. After an MRI, it was confirmed I had a tumour on my cerebellum, which was later diagnosed as a hemangioblastoma. It was successfully removed. Due to my family history of brain tumours, I had a genetic test and I was found to have a gene called Von Hippel Lindau or (VHL). Three months after my tumour was removed, we had a baby boy. He has since also been found to carry the VHL gene.*

*As we wanted to have another child, we spoke to the Familial Cancer Unit at the Women's and Children's Hospital regarding our options. We chose IVF as this was the best option for us to eradicate the VHL gene. We were referred to a Sydney IVF clinic and are now 8 months pregnant with a baby who has a 99% chance of not having the VHL gene. It has been 2 years since my tumour was removed, and I have screening tests every year. The support from my wife, family and friends helped me through the toughest time of my life. I am now fit and healthy and enjoying life to the full.*

Editor's note: This story has been edited for publication purposes.



**Challenges are what make life interesting;  
overcoming them is what makes life meaningful.**  
**Joshua J. Marine**

## Data update from the Familial Cancer Unit

In the past 12 months (July 2005 to June 2006), the FCU counsellors and geneticists have seen 766 people at 540 appointments. (At many of our appointments we see more than one family member).

- New referrals accounted for 549 people at 389 appointments. (Referrals are from GPs and specialist doctors)
- Presymptomatic referrals accounted for 217 people at 151 appointments. (Referrals are from relatives, where the FCU has found a mutation and can test for it in other family members)

Of all new referrals, testing was initiated in 145 families, and 24 mutations were found. This means that out of all the tests performed, the rate of detecting a mutation was about 1 in 6 (or between 16-17%).

## Introducing Dr Nicola Poplawski

A second clinical geneticist has joined the Familial Cancer Unit. Dr Nicola Poplawski trained in New Zealand, Australia, and Canada. She has an MBChB and an MD from the University of Otago, New Zealand. Dr. Poplawski is working part-time with the SA Clinical Genetics Service, dividing her time between the Familial Cancer Unit and the Metabolic Unit.



## Interested in peer support?

The Familial Cancer Unit in conjunction with The Cancer Council South Australia wish to train a small number of volunteers for a peer-to-peer support program for BRCA carriers to be trialled in 2007. Are you:

- a woman with a BRCA1 or BRCA2 mutation?
- comfortable talking with others about decision making around surgery?
- available for two full days of training?

If you are interested and would like to receive more information, please ring Vanessa on 08 8161 7566

# Help us to keep you up-to-date.

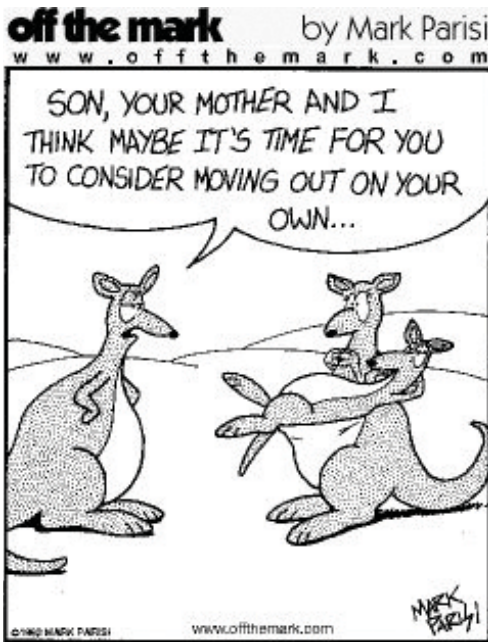
- Have you or any family members changed address?
- Are there any new cancer cases in your family?
- Is there any surgery planned for cancer or preventative reasons?

If you have answered "yes" to any of the above questions please let the Familial Cancer Unit know.

The Familial Cancer Unit  
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## Go Green...

Do you wish to receive GeneTrek electronically and help reduce paper waste? Please email Carolyn Harrington at [charrington@cancersa.org.au](mailto:charrington@cancersa.org.au)



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## Web Watch

The internet is a good way of finding information, but as you may know, sometimes internet information can be unreliable. The aim of web watch is to keep you informed about reliable websites on cancer genetics and related topics. Have a look at the revamped website for The Cancer Council South Australia <[www.cancersa.org.au](http://www.cancersa.org.au)> (familial cancer section currently under construction).

For information on sharing genetic test results with your family, follow the links at "People Living with Cancer" <[www.plwc.org](http://www.plwc.org)> (home page >learning about cancer>genetics>sharing genetic test results with your family). PLWC is a useful and informative web site, but please be aware that the risks of cancer reported here do not necessarily apply in Australia. There are differences in the risks of cancer in different communities and countries. Australian women who carry a BRCA1 or BRCA2 mutation are probably at slightly lower risk of developing breast or ovarian cancer than women who live in North America.

Understanding Gene Testing and complex issues around testing. <[www.cancer.gov/cancertopics/understandingcancer/genetesting](http://www.cancer.gov/cancertopics/understandingcancer/genetesting)> is a reliable and comprehensive site from the National Cancer Institute (United States) discussing topics ranging from what genes are to limitations and benefits of genetic tests, and issues around decision making.

The Cancer Council Australia <[www.cancer.org.au](http://www.cancer.org.au)> has launched the family cancers facility and now hosts The Cancer Genetics Resource Directory <[www.cancergenetics.org.au](http://www.cancergenetics.org.au)>. This valuable national resource directory provides the general Australian public and health professional community with accurate and updated details of a variety of cancer genetics resources. Why not check out the resource directory and spend a few minutes doing the evaluation survey – your input is valued.