



GeneTrek

The newsletter from the **Familial Cancer Unit**

September 2005

Welcome to the second issue of *GeneTrek* for 2005! Please feel free to write in with your thoughts about this newsletter or with any ideas for future articles. It is always great to hear from you.

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Reproductive choices for couples at risk of familial cancer

When people learn that they have inherited a genetic predisposition to cancer, this may be a consideration if they are planning a family. Some individuals or couples may be concerned about the risk of passing on the genetic predisposition and whether there are ways of reducing this risk. There are a number of reproductive options that couples can consider, and these are summarised below. There is no preferred or recommended option. There are usually a number of factors that influence decision-making, and different couples may make different choices.

Some couples may choose no intervention and proceed with starting a family when they are ready,

accepting that their children may or may not inherit the genetic predisposition to cancer. A child who inherits the genetic predisposition may never develop cancer. Couples may also hope that treatment options, or preventive strategies, will continue to improve in the future.

Some couples feel very strongly that they do not want to pass on the genetic predisposition. When the genetic error responsible for the predisposition to cancer has been identified, some couples may wish to conceive a pregnancy naturally and have prenatal testing. This involves testing the developing foetus with a view to termination of the pregnancy if the foetus is shown to have inherited the genetic error.



This newsletter aims to provide

- ◆ **Communication**
- ◆ **Connection**
- ◆ **Current information**

The recommended prenatal test is called Chorionic Villus Sampling (or CVS). This test is usually performed between 10-12 weeks of pregnancy and

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involves using an ultrasound-guided needle to obtain a sample from the developing placenta. Results are usually available two weeks following the procedure and are highly accurate. There is a small risk of miscarriage associated with this procedure. Prenatal testing is accessible through the public health system and is covered by Medicare. There are potential medical and emotional issues associated with prenatal testing which are addressed through supportive counselling.

An alternative option to prenatal testing is Preimplantation Genetic Diagnosis (or PGD). PGD utilises IVF technology to create embryos using the couple's own eggs and sperm. A cell is removed from each embryo and tested for the known gene error. Only embryos found not to have inherited the gene error are selected for transfer to the woman's uterus. If a pregnancy results, it is anticipated that the baby will not have the genetic predisposition.

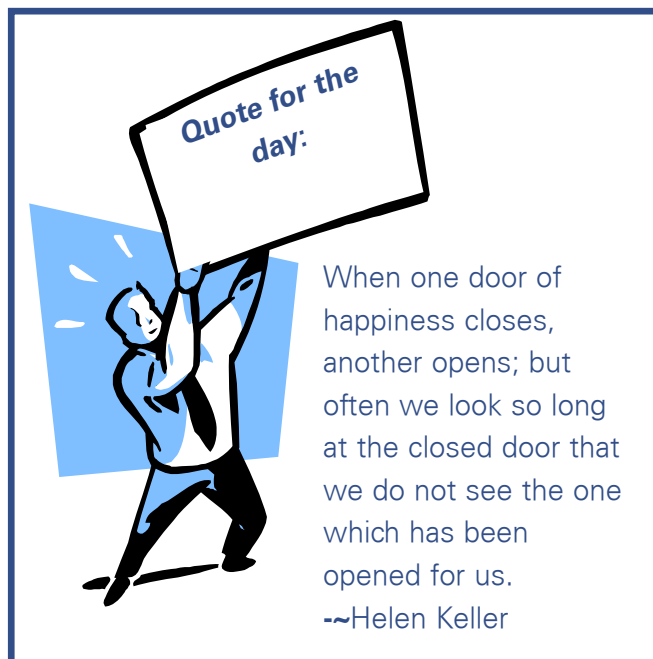
PGD requires a couple who may be otherwise fertile to undergo IVF with the associated medical risks and financial costs, much of which are not covered by Medicare. PGD requires careful work-up for each couple and may not be possible in all cases. There is also no guarantee of achieving a pregnancy following any cycle of treatment. The testing is complex and is not considered as accurate as prenatal testing. For this reason, prenatal testing is usually recommended to confirm the results in any resulting pregnancy.

In some families in which the gene error has not been identified, prenatal testing or PGD may still be possible using a different genetic test called "DNA linkage". This is not widely available, is not as accurate as other methods, and would need to be considered on a case-by-case basis.

There may be couples who do not wish to pass on the genetic predisposition but for various reasons do not wish to use prenatal testing or PGD. Some couples may therefore choose not to have children. Others may wish to explore the possibility of adopting a child. There is also the potential option of using donor eggs, donor sperm or donor embryos (as appropriate) from a person or couple who is unlikely to have the genetic predisposition. With these latter options, there are many ethical, emotional and practical issues that need to be considered and discussed with an appropriate health professional.

There are experienced doctors and counsellors available to discuss all of these reproductive options in detail with couples and to support them throughout their decision making process. It is recommended that these discussions occur well in advance of planning a family.

If you would like further information regarding any of the issues discussed above, please contact one of the genetic counsellors at the **Familial Cancer Unit, Women's and Children's Hospital** on **(08) 8161 6995**.





Relay for Life: be in it!

Each year The Cancer Council holds a Relay for Life event at Santos Stadium, as well as in 7 regional centres around SA. Relay for Life is a team event to raise funds for cancer research, education, prevention and support, and it is a lot of fun. The event is held over 20 hours with bands, late-night movies, competitions and freebies. Teams are formed by friends, colleagues and families. Most teams have a theme and each team has its own tent site and keeps a team member rostered on to walk the track for the whole 20 hours of the event. All team members raise funds to compete for the prize of most successful fundraiser.

The Familial Cancer Unit has had a team in this event since its inception in South Australia in 2001. Graeme Suthers and his team would now like to invite you to join the FCU Genies Team. It is a lot of fun for a worthy cause. Those of us who are clients of the Familial Cancer Unit have a lot to gain from the proceeds of these events. The early identification, treatment and management of our conditions are improved by research, so it is great to be involved in an event from which our families can derive a direct benefit.

I have had the opportunity to walk/run with members of the Familial Cancer Unit teams over the past 3 years. Aside from being required to wear one of Graeme Suthers' funny hats, it is a very useful time to talk generally about the Unit, research and some of the things we do not find time for at clinical appointments. It is also a chance to talk with others in similar situations and realize we are not alone - and not so badly off.

I would encourage everyone to consider being involved in Relay for Life. If you would like to join the Genies Team contact **Jacquie** or **Debbie** at the Familial Cancer Unit on **8161 6995**. If you would like more information about Relay for Life call **Christine Robertson** at The Cancer Council on **8291 4154** or go to the website **www.cancersa.org.au** (go to *Major Fundraising Events* and then *Relay for Life*).

Ian Basey

Ian Basey is a client of the FCU and a member of the organising committee for Relay for Life.



Childhood Leukaemia

Leukaemia is a cancer of the white blood cells. It is one of the most common childhood cancers. As with any cancer, leukaemia is due to the accumulation of genetic errors (mutations) over time. Once a white blood cell accumulates a certain combination of mutations, it begins to grow out of control (leukaemia). The earliest mutations responsible for childhood leukaemia occur in a white blood cell of a developing baby during the earliest stages of pregnancy, well before the mother is even aware that she is pregnant. The majority of the babies who have these early "leukaemia" mutations never develop leukaemia because they are fortunate enough not to develop other mutations in the same white blood cell. But some children do develop other mutations in the same white cell over time, and this eventually causes leukaemia in childhood. It is not known

what factors determine whether a child will develop leukaemia rather than simply having a mutation in an otherwise normal white blood cell.

Sometimes leukaemia occurs in twins. Identical twins are due to a developing embryo (simply a ball of cells at an early stage) splitting into two. Each fragment then develops into a baby. If an early "leukaemia" mutation occurs before the twins split, both of the twins may carry abnormal white blood cells with the mutation. This means that both twins are at increased risk of developing leukemia during childhood. This is what we see in practice: if one twin develops leukaemia, the identical co-twin is very likely to develop leukaemia as well. But note that this is not an inherited tendency. The initial mutation in both twins occurred after conception and was not inherited from either parent. (This is not the situation with non-identical twins.)

Studies of twins have identified when "leukaemia" mutations occur. But it is also clear that the great majority of children with leukaemia, including affected twins, do not have an inherited disorder.

Research: South Australian families teaching the world

The counsellors of the Familial Cancer Unit see 800 clients per year. Sometimes we come across a situation that teaches us something that no-one else knows. One SA family had an inherited tendency to develop bowel and endometrial cancer due to an inherited error in the PMS2 gene. No-one (in the whole world), had been really sure whether mutations in this gene could cause inherited bowel cancer in multiple generations of a family. Detailed evaluation of this local family showed for the first time that mutations in this gene can cause problems in multiple generations. Our report of this family was published in the international journal, *Gastroenterology*, in May 2005.

Another SA family had Cowden syndrome, a disorder due to an inherited error in the PTEN gene. Two sisters in this family developed abnormalities of major blood vessels in the pelvis which have required repeated treatments in hospital. These blood vessel abnormalities are not widely recognized as being a feature of Cowden syndrome, and other patients with this disorder may not be diagnosed until it is too late to provide effective treatment. We wrote a report describing this family's story which was published in the international *Journal of Medical Genetics* in August 2005.

We are grateful to these families for their assistance in taking new information to the international genetics community. If you would like to receive copies of these published reports, or of other research publications from the **Familial Cancer Unit**, please contact us on **(08) 8161 6995**.

Counsellors' Corner

Dear Jacquie

I have inherited a BRCA2 mutation. I have had a bilateral prophylactic mastectomy and prophylactic oophorectomy to reduce my risk of developing cancer of the breast and ovary. What surveillance should I have now?

This can be a tough question to answer, in both medical and emotional terms.

On the medical side of things, the big advantage of prophylactic surgery is that it dramatically reduces the risk of developing cancers of the breast or ovary. As a result of having surgery, your risk of developing these cancers is less than it would be for women in the general population. Some women in this situation (and their doctors) decide not to

have further examinations for breast or ovarian cancer. However, this surgery does not eliminate the possibility of cancer. The risk is certainly reduced, but it is not zero. For this reason, some women prefer to have an annual review by their surgeon or gynaecologist to look for any evidence of problems. But there is no clear medical evidence to support either approach. This is where it can get emotionally tough. On the one hand, you have had major surgery to free yourself of some of the concerns about cancer risk. Avoiding cancer (or detecting it early) can appear to be an endless drain on your emotional resources. On the other hand, cancer (like any illness) can still be a hazard and early diagnosis and treatment can be beneficial.

The decision about cancer surveillance after prophylactic surgery needs to begin with how you feel about your health, the risk of illness in general, and your relationship with your doctor. If you want regular review of your health, then a check of remaining breast tissue and your pelvis by your doctor (GP, breast surgeon, or gynaecologist) every 6-12 months could be part of your general examination. If this causes you anxiety, then you may be more comfortable accepting that the surgery has removed the majority (90% or more) of your risk of these cancers. It really comes down to what you want.

We are more than happy to assist you in working through these issues to find the best fit that suits you.

Clara's Cookbook

All recipes that feature in this newsletter are not only tasty but are also healthy; they have been endorsed by the dietician at The Cancer Council South Australia.

Tabbouleh



Ingredients

- ❖ 1/2 cup cracked wheat (bulgur)
- ❖ 3 cups flat leafed parsley, finely chopped
- ❖ 1/2 cup spring onions, finely chopped
- ❖ 2 ripe tomatoes, seeds removed and finely chopped
- ❖ 1 cup fresh mint leaves (finely chopped)
- ❖ Juice of 2 lemons
- ❖ 2tbs olive oil
- ❖ Coarsely ground black pepper and salt to taste

Cover the cracked wheat with water and let it soak for 10 minutes. Drain well, making sure you squeeze out excess water.

In a large bowl, combine the parsley, spring onions, tomatoes and mint. Stir in the olive oil, lemon juice and salt and pepper to taste. Enjoy!

Editorial responsibility of this newsletter:

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Research at the Familial Cancer Unit

The Familial Cancer Unit is involved in 3 national research projects. These projects investigate aspects of familial breast cancer, bowel cancer and haematological cancers in families where there has been a history of these cancers, usually in a number of individuals over several generations. Participants are asked questions about their health, lifestyle and diet. A small sample of blood or a painless scraping of cheek cells is also taken. If you would like to enquire about enrolling yourself and your family in any of these projects please contact **Meryl Altree**, Senior Research Nurse, Familial Cancer Unit.

Phone: (08) 8161 6821.

Data from the Familial Cancer Unit

Number of South Australian families with known mutations:

Familial Breast Cancer	98
Hereditary Non Polyposis Colorectal Cancer	37
Familial Adenomatous Polyposis	26
Familial Thyroid Cancer	8
Von Hippel-Lindau and Hereditary Paragangliomas	7
Neurofibromatosis Type 2	5
Familial Retinoblastoma	4
Li-Fraumeni	2

Reminder!

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

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

The Familial Cancer Unit

Phone: 8161 6995

Email: famcancer@mail.wch.sa.gov.au



GeneTrek Evaluation

Your feedback is so valuable in the planning of future issues of the GeneTrek newsletter. Please take a few minutes to fill out this evaluation form. Your feedback is most appreciated.

1. What do you most like about the newsletter?

2. What do you least like about the newsletter?

3. Do you have any suggestions for topics that may be discussed in future issues?

Please return evaluation sheet to:

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The first 20 entries receive a free Daffodil Day bear from The Cancer Council. Children of all ages love them!
(Please supply your contact details)

Name:

Address:
