



Gene Trek

Familial Cancer Unit newsletter

May 2010



From the editor

Dear Readers

Welcome to the first edition of Gene Trek for 2010. We have two feature articles in this edition. The first provides information about the impact of recent changes to the Federal Privacy Act. The second takes a look at the genetics behind cancer and how familial cancer differs from non-familial cancer. In our Counsellors' page, there is a list of some great resources and support options for people who undergo surgery to reduce their cancer risk. We take a close look at surveillance letters in our data update, and there is a yummy, but healthy apricot strudel recipe for winter.

If you have any feedback about this edition or any suggestions for future topics, I'd love to hear from you.

Kind regards

Kirsty Stallard

Cancer Genetics Education
Project Officer
Cancer Council SA
PO Box 929
Unley BC South Australia 5061
t 08 8291 4269
f 08 8291 4268
genetics@cancersa.org.au



Staff update

Farewell Kate Ryan

After just over two years of service as a Genetic Counsellor at the Familial Cancer Unit, Kate Ryan has accepted a Genetic Counselling position in another Unit of the South Australian Clinical Genetics Service, and will have other responsibilities. We thank Kate for her contributions and wish her well in her new role.

In this issue

Confidentiality and recent changes to the Federal Privacy Act by Dr Graeme Suthers

The genetics behind cancer by Dr Graeme Suthers & Dr Nicola Poplawski

Your place—Karyle's story

Counsellors' page—information and support for people having surgery to reduce cancer risk

Research updates—updates from kConFab and ACCFS studies

Data update from the Familial Cancer Unit—surveillance letters under the spotlight

Healthy eating—Apricot strudel

Resource update—Cancer Council SA, websites, support groups

Confidentiality and recent changes to the Federal Privacy Act

Dr Graeme Suthers, Clinical Geneticist and Head, Familial Cancer Unit, South Australian Clinical Genetics Service, Women's & Children's Hospital, North Adelaide, South Australia

Patients often need to reveal intimate details about their personal health in order for their healthcare practitioners (such as doctors, nurses and counsellors) to provide appropriate and effective advice or treatment. This personal information is regarded as confidential, that is, it is not revealed or made available to anyone outside the immediate professional team caring for a patient. As confidentiality is a fundamental aspect of healthcare, stringent professional and legal obligations are placed on all healthcare practitioners in this regard.

These obligations are laid down in professional codes of conduct and in state and federal legislation, including the Federal Privacy Act. To a large extent, these various codes are in agreement but there are some important distinctions. In particular, healthcare practitioners in private practice are subject to federal privacy legislation, while healthcare practitioners in the public sector (including the Familial Cancer Unit) are only subject to state privacy legislation. This difference has important implications and is discussed below.

Confidentiality in familial disorders

The limits of confidentiality can become more complex when considering a familial disorder. In this situation personal health information about relatives may have medical implications for a patient. In order to make an assessment, the healthcare practitioner needs to obtain and record personal health information about their patient's relatives. However, the relative in question is usually not present at the appointment to give consent for their information to be obtained or held by the healthcare practitioner.

Federal privacy laws allow healthcare practitioners to obtain and record such personal health information about their patient's relatives, provided that the information collected is relevant for the patient's care and that it is only used for that purpose.

Relatives at risk

On the flip side, healthcare practitioners also need to consider that information about the patient can also

carry healthcare implications for relatives. For example, if a patient is known to carry a mutation in a high risk cancer gene, their relatives may carry the same mutation and be at significantly increased risk of developing cancer. Having access to such information may be very helpful to the relatives and their doctors by enabling them to reduce the risk or severity of disease through screening and preventive approaches, or in reducing the need for unnecessary investigations by excluding them as carriers. This raises the question: who is responsible for telling the relatives about this important yet personal health information?

Informing relatives of genetic risk

A healthcare practitioner is responsible for making the diagnosis of a familial disorder in a patient. To make such a diagnosis, the practitioner must have obtained, recorded, and interpreted information about a patient's family history of disease. As noted above, there is legal provision for the handling of this information.

The practitioner must also advise the patient of this diagnosis and of the potential relevance of this diagnosis for relatives. As with any professional advice, it is prudent to have this advice documented in writing. It is for this reason that the Familial Cancer Unit provides written advice to every client. In addition, there is legal precedent for practitioners being held responsible for failing to advise patients about the implications of a diagnosis for their relatives.

As a matter of course, most patients will provide this information to their relatives. However, some patients find sharing personal health information with relatives particularly difficult. To make it easier, a healthcare practitioner may assist in contacting relatives with the consent of the patient. However, it is worth noting that the healthcare practitioner is under no legal obligation to do so. The Familial Cancer Unit offers such assistance in families with an identified mutation in a high risk cancer-gene. In our experience we have found a combination of patient-to-relative communication (often face to face) and practitioner-to-relative communication (usually by mail) is quite effective in sharing such important health information in a family.

Patient refusal to share genetic risk information with their relatives

There is one situation that has not been covered in this discussion. What if the patient refuses to provide information about genetic risk to relatives, and refuses to allow the practitioner to provide this information?

In the past, Australian legislation did not allow healthcare practitioners to release confidential information to

relatives without the consent of the patient, even if such information might be lifesaving for the relatives. This situation has now changed.

Recent changes to the Federal Privacy Act

The Federal Privacy Act has been amended to allow healthcare practitioners to advise relatives about the risk of a familial disorder even if the patient has refused to allow release of this information. It is important to recognise that such release flies in the face of a strong emphasis on the confidentiality of medical information in the past. The amendments to the Privacy Act clearly state that such a release of information against the wishes of a patient can only occur in certain situations:

- there is a serious threat to health of a genetic relative
- providing confidential information to the relative is necessary to reduce that threat, and
- other approaches to release the information with consent have been exhausted.

The NHMRC has released Guidelines for practitioners who may need to consider releasing such information against the wishes of a patient (see <http://www.privacy.gov.au/materials/types/guidelines/view/7015>). It is important to note that these Guidelines are not merely 'good advice' about disclosure. A practitioner must comply with these Guidelines to maintain compliance with this section of the Privacy Act. It is anticipated that this provision of the Privacy Act will be utilised rarely.

This amendment to the Privacy Act applies only to practitioners in the private sector. Each State will need to amend its own privacy legislation to allow practitioners in the public sector (such as the Familial Cancer Unit) to release information without consent. This issue has been raised with SA Health, but we do not know how long it may take to have the local legislation amended.

If you have further questions about privacy and health-related information, see <http://www.privacy.gov.au/law/act/genetic> Please feel free to contact us on 08 8161 6995 or at Famcancer41300@health.sa.gov.au if you have other concerns.

The genetics behind cancer

Dr Graeme Suthers and **Dr Nicola Poplawski**

Clinical Geneticists

Familial Cancer Unit

South Australian Clinical Genetics Service

Women's & Children's Hospital

North Adelaide, South Australia

Cancer is common in Australia. 1 in 3 people in our community will have experienced at least one form of cancer by the age of 70. By 85 years of age this chance increases to 1 in 2. In fact, if a person does not die of something else first, the ultimate risk of developing cancer at some time during a person's life is 100%. The reason for this lies in our genes, and the errors that arise when we copy our genes.

It all begins at conception, when a sperm fertilises an egg

When a sperm fertilises an egg the first cell that will go on to make a person is created. That first cell contains two copies of each of the 20,000 genes or 'genetic recipes' that make up a person's genetic code (their DNA code). Genes provide the instructions needed for an egg to develop into a baby, the baby into a child and the child into an adult. By the time a person reaches adulthood they have approximately one hundred million million cells, and each of those cells contains a copy of the original set of genes.

Our genetic code is very long—about three billion 'letters' long. Each cell has two copies of the genetic code, which means each cell has about six billion letters of code. As a cell has a limited life span, the body is constantly making new cells. It is important that a new cell contains the same genetic code as the cell it replaces, so it can function properly. To do this, the old cell must make an identical copy of its DNA.

Although DNA copying is very accurate, a small number of copying errors arise each time a new cell is made, purely because of the enormous length of genetic code that must be copied. So, as we get older we gradually accumulate genetic 'spelling errors' in our genes. This is a normal phenomenon and occurs in every cell, in every person. This is sometimes referred to as 'acquired genetic rust'; or a more technical term is 'acquired genetic mutations'.

Most cancer is associated with acquired genetic mutations

If the human body is to work properly, it is important that the growth of its cells be tightly controlled. Each normal cell contains many genes which control the growth of the cell. The presence of many growth-control genes provides a back-up mechanism in the event that one of the growth control genes fails to work correctly. But, as we get older, the progressive accumulation of acquired mutations in a cell may eventually interfere with enough of the growth-control genes within that cell for it to begin to grow out of control. We refer to this as sporadic cancer. This explains why cancer is common and why it usually occurs in later life.

People vary in their risk of developing cancer by a certain age, as the rate at which mutations are acquired is different for every person. Although we cannot predict how quickly a particular person will accumulate mutations, we do know that a person accumulates more and more mutations as they get older. We also know that the rate at which a person accumulates mutations is influenced by

- exposure to environmental agents (like tobacco smoke, asbestos dust, UV light, certain viral infections) and
- lifestyle (like poor diet, physical inactivity, obesity, alcohol consumption) and
- chance ('bad luck').

Most cancers develop because of a combination of all of these factors, as well as other factors we do not currently understand.

In sporadic cancer, only the cancer cells have the acquired cancer-causing mutations. In hereditary cancer the situation is different—every cell in the person's body has a mutation in a specific cancer-predisposition gene (an inherited genetic mutation) and, in addition, the cancer cells also have additional acquired cancer-causing mutations.

Inherited genetic mutations and cancer predisposition

Occasionally a person can inherit a genetic mutation in one of the genes that control cell growth. The mutation was present in the egg or sperm from which the person developed, and was copied into every cell of a person's body as it grew and developed. The presence of this cancer-predisposing mutation in every cell places the person at high risk of developing cancer at a young age. This is because an individual cell needs to accumulate fewer additional acquired mutations in growth-control genes to become a cancer.

People with an inherited mutation in a growth-control gene are at increased risk of developing cancer compared to the general population. These people may have a family history of the same or related cancers, and they may also pass this predisposition to cancer on to their children. This inherited tendency to develop cancer is known as familial cancer. The hallmarks of familial cancer include a family history of close relatives having the same type of cancer, a diagnosis of cancer at a younger age than usual, and a person developing more than one primary cancer.

Many of you would already be acutely aware of the incidence of cancer or indeed, the presence of a mutation in a cancer-risk gene in your family. Knowing this provides a golden opportunity to intervene and

reduce the impact of cancer for family members through targeted surveillance programs and other risk reducing strategies.

If you wish to speak to a genetic counsellor about the cancer in your family please call the Familial Cancer Unit on 08 8161 6995.

Research update

Update from the Australasian Colorectal Cancer Family Study

The Australasian Colorectal Cancer Family Study (ACCFs) was founded by Australian colorectal pathologist Professor Jeremy Jass in the late 1990's. Since the study began, more than 10,000 Australians and New Zealanders from 1,800 families have participated in the study. The aim of the study is to provide a resource for colorectal cancer researchers to enable them to better understand the role genes and the environment play in bowel cancer.

The Study has recently secured a further three years funding from the National Institutes of Health (NIH) USA as part of the worldwide Colon Cancer Family Registry (Colon CFR). This new funding is enabling ACCFS to invite more families and relatives to participate in the study. It is also helping researchers maintain contact with participants who have previously enrolled so their information can be updated.

Recruitment of suitable high risk families occurs through familial cancer clinics and the community. Such families have three or more cases of bowel cancer. In South Australia, an ACCFS research nurse is based at the Familial Cancer Unit. If you wish to find out more about the study or recruitment process please contact Louise Jaensch on 08 8161 6995 or Louise.Jaensch@health.sa.gov.au

Louise Jaensch, ACCFS Research Nurse, Familial Cancer Unit, North Adelaide, South Australia

Update from the kConFab research project

The Kathleen Cunningham Foundation Consortium for research into familial breast cancer (kConFab) was established in 1997. Since its inception, more than 1,400 families have donated tissue and data to the study. This resource is accessed by approved researchers, enabling them to discover more about the familial aspects of breast cancer, and the implications of BRCA1 and BRCA2 mutations.

Recently, kConFab changed their criteria for the types of tissue they would like to collect from families already

enrolled in the study. From these families, they are now interested in collecting

- prophylactic breast tissue
- ALL tumour tissue (including breast, ovarian, bowel, pancreatic, prostate).

If you have participated in the kConFab research project and are interested in donating a small sample of tissue collected at surgery, please contact Kerry Phillips on 08 8161 6124 or Kerry.Phillips@health.sa.gov.au

I would like to extend a big thank you to all participants. Your contribution will benefit current and future generations.

Kerry Phillips, kConFab Research Nurse, Familial Cancer Unit, North Adelaide, South Australia

Your place

My life changed the day my sister was diagnosed with aggressive breast cancer at the age of 33. To our family's knowledge, she was the first. My initial reaction was "I'm going to lose my sister...", but we are a close family who pulled together, then our fight for her life began.

After six months of living in a vacuum, the rounds of chemo and radiotherapy treatment finally came to an end and life began to return to normal—almost. For me, however, my journey continued. With exceptionally lumpy breasts that were almost a size DD cup, and already experiencing some concern, I became increasingly more paranoid. Checks went from one or two a month to almost weekly and visits to the doctor became six-monthly. It was a fear I lived with day and night.

Then in 2005, Terry received a letter from the Familial Cancer Unit recommending that she book an appointment to revisit the possibility of a genetic link. After months of waiting on eggshells, the result came through—Terry had BRCA1.

My paranoia now increased ten-fold. Checking my breasts became a daily ritual, often carried out several times between waking and sleeping, and visits to the doctor were now quarterly. Looking back, it is almost as though I subconsciously knew there was a reason for me to be worried, as January 2006 brought the news that I too had BRCA1.

My course of action was determined. With no regrets, I had removal of my ovaries and fallopian tubes (as well as a hysterectomy) and then six months later I had a mastectomy and reconstruction. Many people have often said, "You are so brave." I don't see it as bravery, but as meeting life's challenges head on, then living life to the fullest.

My family's cancer experience and facing my own mortality has helped me realise that life is too short to always live in one's own comfort zone. As a result, I changed careers and am now enjoying being a professional photographer. One of my aims as a photographer is to provide men and women who go through this life changing experience with the opportunity to have tasteful before and after black and white portraits taken, with part of the proceeds being donated towards genetic cancer research.

Had I not travelled down the path of discovering I carried the BRCA1 gene, I would not be the person I am today. Discovering I had BRCA1 was the best thing that ever happened.

Karyle

(Editor's note: Examples of Karyle's photography can be viewed on her website www.karylesphotography.com.au)

Counsellors' page

The genetic counsellors from the Familial Cancer Unit



Left to right: Jacquie Armstrong, Debbie Trott, Sally Russell, Vanessa Huntley

What resources are available to women considering a prophylactic or risk reducing mastectomy?

Deciding whether risk reducing breast surgery is right for an individual is very complicated and requires women to weigh up many factors before reaching a conclusion. This takes time and can also feel overwhelming. There are a number of places that women can obtain further information and support to help them in making this decision.

Specialists

Breast surgeons are the specialists in this area and can provide you with detailed information about the medical procedures involved in mastectomy and breast reconstruction based on your health and body type. Usually this will also involve speaking to a plastic surgeon for more detailed information about breast reconstruction.

Genetic Counsellors

Genetic counsellors are available to talk to women about the reduction in cancer risk achieved by this type of surgery. They can assist women to identify the most important physical and emotional implications to them from such surgery and to provide support to individuals or couples during the decision-making process. Support may also be important at the time of surgery and during the recovery process. A psychologist may also be an appropriate person to provide support around decision making.

Books, DVD's and pamphlets

This is a relatively new area of medicine and the resources available on this specific topic are limited. Breast surgeons, plastic surgeons and the Familial Cancer Unit each have different resources available to women. It can be helpful to access a number of different types to assist in your decision.

Talking to another woman (peer support)

Many women find it helpful to talk to another woman who has had risk reducing breast surgery. Sometimes this may help a woman identify potential issues not thought of before or allow questions that only a woman who has 'been there' could answer. Most breast and plastic surgeons are able to put their patients in touch with a volunteer if a woman requests this. Volunteers are also available through the Cancer Council Cancer Connect service. Call **Cancer Council Helpline 13 11 20** to make an appointment.

Internet

The internet can be a useful resource for women looking for information about risk reducing breast surgery. There are an increasing number of websites available in Australia and overseas devoted to women at an increased risk of developing breast cancer. Some caution is recommended though as most of these websites are unmonitored. This means that some information may not be accurate or applicable to the Australian health care system. A reliable starting point is the National Breast and Ovarian Cancer Centre (NBOCC) website www.nbocc.org.au

Jacquie Armstrong

It can take time to adjust to living with an increased risk of cancer. The important people in your life can also be affected. Issues can arise weeks, months or even years later. If there are any issues that you would like to discuss in more detail, for example surveillance recommendations, risk reducing surgery or exploring your feelings, please feel free to contact us at any stage on 08 8161 6995.

Data update from the Familial Cancer Unit

Surveillance letters under the spotlight

Living with the knowledge you have an increased risk of developing cancer can be difficult. For some people there may be options available to them to manage their cancer risk. Such options may include approaches to reduce cancer risk or cancer surveillance strategies to detect cancer early. The risks and benefits associated with both options should be discussed with the health professionals managing your care.

Reducing cancer risk may involve relatively simple lifestyle strategies such as healthy eating, not smoking, getting regular exercise, reducing alcohol intake and taking care in the sun. At the other end of the scale, reducing cancer risk may involve the use of surgery to remove at risk tissue, for example prophylactic removal of breast tissue in a mastectomy.

It is well established that early detection of cancer gives a person the best chance of successful treatment. Cancer surveillance strategies may range from self monitoring for changes or symptoms (e.g. self breast checks) to cancer screening using medical imaging techniques (e.g. mammography), procedures (e.g. colonoscopy), or pathology testing (e.g. Faecal Occult Blood Testing (FOBT)).

To be effective, cancer surveillance should be carried out regularly. The suggested interval between checks will be determined by current evidence and the healthcare professionals looking after you. It will also be determined by what you are comfortable with. To help with reminding you when your surveillance checks are due, the Familial Cancer Unit can send you a surveillance reminder letter by mail.

Since the year 2000, the number of surveillance letters we send has increased dramatically (see figure). Up until the year 2002, less than 50 surveillance letters were sent out annually. In 2009, nearly 1600 surveillance letters were sent out; more than a 15-fold increase in 10 years.

In addition to this, the types of surveillance reminders we send out has broadened significantly. Currently, we send out surveillance reminder letters to people from families at high risk of

- familial breast/ovarian cancer
- familial bowel cancer (including HNPCC, FAP and other polyposis syndromes)
- Multiple Endocrine Neoplasia types 1 and 2
- Neurofibromatosis type 2
- Peutz-Jeghers syndrome
- Li-Fraumeni syndrome
- von Hippel-Lindau disease
- Hereditary diffuse gastric cancer
- familial pancreatic cancer
- Cowden syndrome
- paraganglioma
- thyroid cancer.

Being reminded when your cancer surveillance is due can trigger many emotions. However, some of the benefits of receiving surveillance letters may include:

- receiving updates to recommended surveillance as new evidence emerges

- having your surveillance results confidentially stored in one location for your future reference
- surveillance results can be sent to the doctors managing your care.

If you wish to receive surveillance updates, as well as the other associated benefits, please contact the Familial Cancer Unit on 08 8161 6995 or at Famcancer41300@health.sa.gov.au

Resources

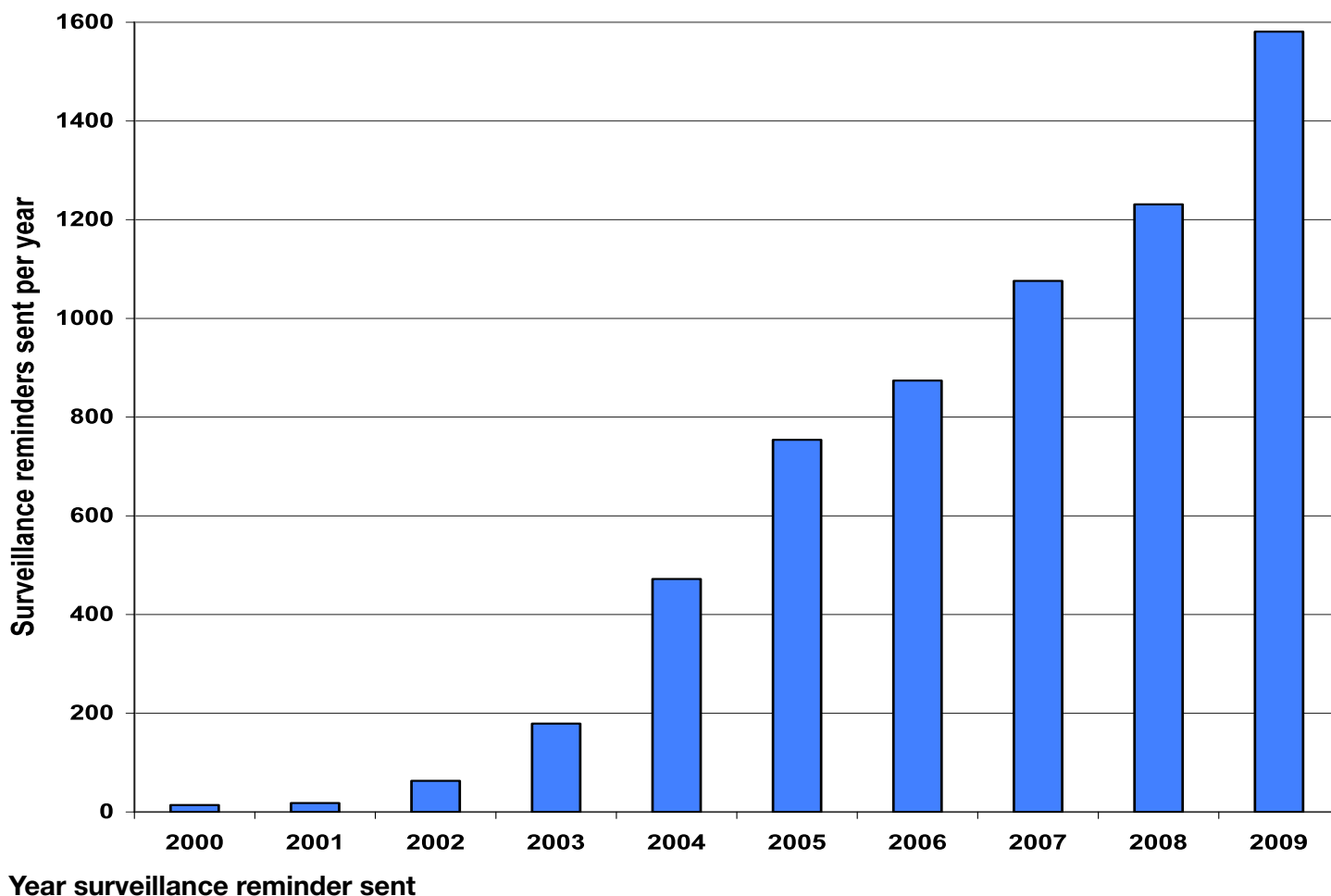
On the web

The **Centre for Genetics Education** has a new look website. If you are looking for reliable information about genetics, inheritance and familial cancer, their factsheets are a great place to start <http://www.genetics.com.au/home.asp>

Support

Access information and support over the phone by calling **Cancer Council Helpline 13 11 20**. Trained oncology nurse counsellors are just a phone call away between the hours of 8.30am to 8.00pm Monday to Friday.

Figure: Surveillance updates sent per year from 2000 to 2009



NEW cancer prevention pamphlet

Cancer Council SA has a new pamphlet. **Cut your cancer risk** provides lifestyle advice to help you reduce your cancer risk. It is available from **Cancer Council Helpline 13 11 20**.



Healthy eating

Apricot Strudel



15 minutes preparation + 25 minutes cooking
7 serves of fruit in this recipe

800g can apricots, drained
1 tablespoon honey
1 teaspoon cinnamon
½ cup dates, chopped
4 sheets filo pastry (27cm x 47cm)
Olive or canola oil spray
2 tablespoons almonds, flaked

Preheat oven to 200°C. Combine apricots, honey, cinnamon and dates in a small bowl. Lightly spray pastry with oil. Lay pastry sheets on top of each other then spoon apricot mixture along the centre length. Fold pastry to encase, tucking neatly into a roll. Spray top with oil and sprinkle with almond flakes.

Bake for 20–25 minutes until golden brown and crispy.

Serves 6.

Hint

Always choose fruit canned in 'natural' juice or pie apples as these do not have 'added sugar'.

Variation

Canned peaches or apples could replace the apricots.

© Healthy Food Fast, State of Western Australia, 2008. Reproduced with permission.

Go green

Would you like to receive Gene Trek electronically? If so, please email your request to the Familial Cancer Unit at Famcancer41300@health.sa.gov.au

Let us know

- Have you or any of your family members changed address?
- Has there been a new cancer diagnosis in your family?
- Is there any surgery planned for cancer or risk reducing reasons?

Have you answered 'yes' to any of the above questions? To ensure we can provide you with the best possible service, please let the Familial Cancer Unit know of these changes on 08 8161 6995 or Famcancer41300@health.sa.gov.au

Editorial responsibility for Gene Trek is taken by:

Dr Graeme Suthers, Head of the Familial Cancer Unit, SA Pathology, Women's and Children's Hospital, 72 King William Street, North Adelaide SA 5006

Printed on ENVI carbon neutral paper