



Cancer Genetics Gazette

A newsletter for specialist clinicians from the Familial Cancer Unit

July 2010

Issue 9



Welcome to the 1st edition of the Cancer Genetics Gazette for 2010. We have 2 feature articles in this edition. The first article, by Dr Graeme Suthers, provides information about the impact of recent changes to the Federal Privacy Act. The second article shows you the who, why and how of DNA banking when a patient has cancer and Dr Nicola Poplawski provides us with an example of how DNA storage can be beneficial to families with familial cancer. We hope you find this edition useful and informative.

Kind regards

Kirsty Stallard

Cancer Genetics Education Project Officer

Cancer Council SA

e genetics@cancersa.org.au

Confidentiality and recent changes to the Commonwealth's *Privacy Act 1988*

Dr Graeme Suthers PhD FRACP FRCPA

Clinical Geneticist and Head of Unit

Familial Cancer Unit

Women's and Children's Hospital

North Adelaide, South Australia

Patients often need to reveal intimate details about their personal health in order for their health care 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 health care, stringent professional and legal obligations are placed on all health care practitioners in this regard.

These obligations are laid down in professional codes of conduct and in state and federal legislation, including the Commonwealth's *Privacy Act 1988*. To a large extent, these various codes are in agreement but there are

some important distinctions. In particular, health care practitioners in private practice are subject to federal privacy legislation, while health care 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 health care 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 health care practitioner.

Federal privacy laws allow health care 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, health care practitioners also need to consider that information about the patient can also carry health care 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 health care 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 health care practitioner may assist in contacting relatives with the consent of the patient. However, it is worth noting that the health care 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 health care 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 Privacy Act

The Privacy Act has been amended to allow health care 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.

DNA banking: storing DNA samples from patients with cancer

There are many patients with cancer in whom genetic testing of cancer predisposition genes is indicated. However, referral to the Familial Cancer Unit for a discussion about genetic testing is not always appropriate. For example, the patient may be receiving palliative care and wish to focus on other issues; the patient may not want to know their own genetic status; the patient may have another medical condition (e.g. depression) which means genetic testing is potentially harmful to them. In these situations DNA can be stored so genetic testing can occur at a later time, or after their death, if this is appropriate.

Which patients with cancer should consider banking DNA?

DNA banking should be considered if the patient has the following family history:

- two or more close relatives (same side of family) with the same or related cancers
- one or more close relatives (same side of family) with the same or related cancers and one of the affected individuals experienced cancer under the age of 40 years
- any relative with cancer under the age of 30 years
- any relative with two primary cancers of the same or related type(s)
- any relative with epithelial ovarian cancer, regardless of age.

Why bank DNA samples?

- DNA studies on fixed tumour tissue are not usually possible due to the low yield and poor quality of the extracted DNA
- lymphocyte DNA from an affected family member is often essential to perform genetic studies in a family with a familial disorder
- a DNA sample can be used before or after a patient's death but only after appropriate written consent is obtained
- giving a blood sample for future genetic studies is a very practical way for a dying patient to provide their family with the option of genetic testing in the future
- testing may make a genetic diagnosis in the family providing at-risk relatives with the option to clarify their risk.

How to bank a DNA sample

- Collect 20ml of blood in EDTA and send with request form to either

Molecular Pathology Laboratory	Genetic Pathology Laboratory
IMVS	Flinders Medical Centre
Frome Road	Flinders Drive
Adelaide SA 5000	Bedford Park SA 5042

- Please include a brief note about the patient's personal and family history of cancer.
- If a relative is seeking genetic counselling, please refer them to the Familial Cancer Unit, Women's & Children's Hospital, 72 King William Rd, North Adelaide SA 5006, telephone 08 8161 6995.

The benefits of DNA storage: a case report

Dr Nicola Poplawski MBChB MD
Clinical Geneticist, Familial Cancer Unit
Women's & Children's Hospital
North Adelaide, South Australia

Sue experienced breast cancer at age 57. Two years later she was diagnosed with primary peritoneal cancer. Review of her histology confirmed the tumour was an

invasive serous papillary adenocarcinoma. She reported a family history of cancer; a brother with prostate cancer at age 64, her mother with breast cancer in her 40s and a maternal aunt with breast cancer at 82. This was sufficient to place the family in a high risk category for both breast and ovarian cancer.

Sue was ambivalent about genetic testing. She had just completed chemotherapy and was looking forward to a period of 'normal life'. In addition, her daughters had differing opinions about her having a genetic test, and she knew a family who had struggled to come to terms with the results of genetic testing for familial colorectal cancer. After long consideration she declined genetic testing.

Six months later, a family member contacted the Familial Cancer Unit (FCU) to inform us that Sue had entered a palliative phase of treatment. Sue was offered immediate genetic testing which she declined, as she and her family wanted to focus on 'the important things in life'. Instead Sue elected to store a DNA sample as one of her daughters wanted to have the option of pursuing genetic testing in the future. She died two months after banking her DNA.

Four years after Sue's death, one of her daughters contacted the FCU to discuss genetic testing. It had taken some time for the family to recover from her death, and they had only recently felt ready to pursue further information about genetics. A family appointment was held with Sue's partner and her two daughters. At the end of the appointment her partner provided written consent for BRCA1 and BRCA2 testing on her stored sample.

Genetic testing of Sue's DNA identified a pathogenic BRCA1 mutation. Her daughters are now considering pre-symptomatic testing to clarify their cancer risk. If they have inherited the BRCA1 mutation, they can explore both cancer surveillance options and risk reducing surgical approaches, to manage their breast and ovarian cancer risk. If they have not inherited the BRCA1 mutation, their personal risk of breast and ovarian cancer is dramatically reduced and they do not require cancer surveillance on the basis of their family history.

If Sue had not decided to store a DNA sample, her daughter's and other at-risk relatives would not have the opportunity to clarify their personal risk, until another family member experienced breast or ovarian cancer.

Editor's note: Client's name changed for publication purposes.

Jewels from the Journals

Contralateral breast cancer risk in the BRCA1 and BRCA2 mutation carriers

Graeser et al, *J Clin Oncol* 2009;27:5887–5892 (and editorial 5862-5864)

This paper demonstrates that BRCA1/2 mutation carriers who experience a first breast cancer have a significant risk of contralateral breast cancer. The risk of a second primary was higher for BRCA1 carriers compared to BRCA2 carriers, and the risk was inversely correlated with the age at first breast cancer diagnosis. The 25-year cumulative risk of a second primary was highest for BRCA1 carrier women who were <40 years at the time of their first diagnosis (62.9%). These findings have significant implications for the management of BRCA1/2 carriers with a first breast cancer.

Expanded extra colonic tumour spectrum in MUTYH-associated polyposis

Vogt et al, *Gastroenterol* 2009;137:1976–1985 (and editorial 1883-1886)

This study of a large cohort of patients with multiple adenomatous polyposis associated with mutations in the MUTYH gene has confirmed the increased risk of gastric and duodenal polyposis, as well as an elevated risk of duodenal cancer (lifetime risk 4%). The study also demonstrated an increased risk of a number of extra-intestinal cancers. However, the spectrum of cancers and the relatively advanced age at onset means additional surveillance measures, apart from regular upper GI endoscopy are unlikely to be helpful in individuals with MUTYH-associated polyposis.

Evaluating women with ovarian cancer for BRCA1 and BRCA2 mutations. Missed opportunities.

Meyer et al. *Obstet Gynecol* 2010;115:945–952

This US-based study confirmed that women with epithelial ovarian cancer continue to be under referred for consideration of BRCA1 or BRCA2 mutation testing (less than 50% of substantial-risk patients were referred). The referral rate for women presenting with recurrent disease was lower than for women with newly diagnosed disease. [Note: the South Australian Familial Cancer Unit offers BRCA1/2 genetic testing to all women with epithelial ovarian cancer, independent of the family history of breast and ovarian cancer. Population based studies indicate ~15% of women with epithelial ovarian cancer will have a mutation identified].

Resource update

New online risk assessment tool

The National Breast and Ovarian Cancer Centre (NBOCC) have released a new online tool for health professionals to assess familial risk of breast and ovarian cancer www.nbocc.org.au/fraboc

Cancer resources

To access a range of free cancer resources call Cancer Council Helpline 13 11 20.

Go green

If you prefer to receive the Cancer Genetics Gazette electronically please email your request to genetics@cancersa.org.au

Feedback?

If you have any feedback from this edition or suggestions for future topics please email Kirsty Stallard, Cancer Genetics Education Project Officer at genetics@cancersa.org.au

Editorial responsibility for this newsletter is taken by:

Dr Graeme Suthers
Head, Familial Cancer Unit
SA Pathology
Women's & Children's Hospital
72 King William Rd
North Adelaide SA 5006

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