



The Gene Pool

The Cancer Genetics Newsletter
for GPs

May 2010



From the editor

Dear Readers

Welcome to the first edition of The Gene Pool for 2010. This newsletter provides information and updates for GPs about familial cancer research and practice. If you would like to request a topic or have any questions or feedback please feel free to contact me.

Kind regards

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

Dr Graeme Suthers, Clinical Geneticist and Head of the 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 Commonwealth's *Privacy Act 1988*. 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

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 Privacy Act

The 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.

Why we don't test unaffected patients

Dr Nicola Poplawski, Clinical Geneticist, Familial Cancer Unit, South Australian Clinical Genetics Service, Women's & Children's Hospital, North Adelaide, South Australia

In the Familial Cancer Clinic most of the clients we see come from families with a cancer history suggestive of an inherited cancer predisposition. In some of these families the pattern of cancers gives us an indication of the specific gene or genes which may explain the family history. This subgroup of families is offered genetic testing in the hope that we can identify the mutation in the family—a process we call a 'mutation search'. If a mutation search is appropriate for a particular family, a common question we encounter is "why do you have to test someone with cancer first?"

A mutation search is not straightforward. A useful analogy is to consider each gene to be a recipe in a large library of recipe books. If after assessing the family history (pedigree) we suspect there is a spelling mistake (pathogenic mutation) in one of these recipes (genes), and we know which recipe to look at (e.g. BRCA1), and we know the recipe's location in the library (chromosome 17), we can take this recipe 'off the bookshelf' and use a range of testing techniques to try and identify a specific spelling error in that recipe. However, no genetic testing technique is 100% sensitive. If a mutation search does not identify an error we are left not knowing if we missed the relevant error, or if the error is in a different (untested) recipe somewhere else in the library, or if the person was not an error carrier in the first place.

A mutation search is expensive. As public health care resources are finite, a mutation search is targeted at individuals in whom there is a reasonable chance of detecting a mutation. However, because we do not understand all the genes and genetic mechanisms that cause an inherited predisposition to develop cancer, most of the time we expect the result of the mutation search to be normal or uninformative; i.e. a mutation is not identified in the gene or genes tested. For example, about 15% of familial breast cancer is explained by mutations in the two genes we commonly test (BRCA1 and BRCA2); in the remaining 85% of families the underlying gene is not known. In the UK the testing threshold is usually set at a 20% or greater chance of detecting a pathogenic mutation; in Australia and North America a 10% threshold is generally accepted. This means a pathogenic mutation is not found in the majority of samples tested.

Most cancer predispositions are inherited in a dominant pattern—if the chance of detecting a mutation in a cancer affected relative is 10%, testing an unaffected first-degree relative reduces the chance of detecting a mutation by 50% (to 5%), and testing an unaffected second-degree relative reduces this chance by 75% (to 2.5%).

Thus, if an unaffected relative is tested we have a greater chance of a 'normal' result (i.e. not detecting a mutation) compared to testing an affected relative. Also, if the mutation search is 'normal', we are faced with a dilemma: the 'normal' result could mean that they have not inherited the family cancer-predisposing mutation (a true negative), or it could mean that we have failed to look in the right gene (a false negative), or it could mean that there is no familial predisposition to cancer (i.e. the family history is 'bad luck') and we cannot tell these three possibilities apart.

In short, using our current genetic testing technologies there are significant limitations in our ability to interpret the result of a mutation search in unaffected relatives. Testing unaffected relatives is also a poor use of our finite health care funding.

There are occasional situations where it may be appropriate to make an exception to the general rule of 'doing the mutation search in a cancer-affected family member':

- testing the intervening but unaffected relative of two individuals who have experienced cancer (e.g. where a mother experienced young onset breast cancer, her son is unaffected, and his daughter also experienced breast cancer, it may be appropriate to test the son who is the intervening relative)
- testing an unaffected person at risk of specific rare syndromes i.e. von Hippel-Lindau syndrome or MEN2, in which the false negative rate for genetic testing is known to be very low. Even in this setting, it is preferable to document the family's mutation by testing an affected relative first.

Once an inherited mutation has been identified in the family the situation is quite different. Because we know which gene to test, as well as the specific mutation to test for, any family member who is at risk of inheriting the mutation can be tested. Testing can be offered to family members who have experienced cancer (confirmation testing) as well as to family members who have not experienced cancer (presymptomatic testing).

Resource update

NEW Online learning module

An online learning module for familial cancer is now available on the Think GP website. Learn how to collect an appropriate family history, perform cancer risk assessment, manage cancer risk through screening and prevention and refer patients to a Familial Cancer Service. CPD points available www.thinkgp.com.au

NEW Online risk assessment tool

The National Breast and Ovarian Cancer Centre 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**.

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