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Editorial

Genetic Risk Counselling for Breast Cancer Families

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INTRODUCTION

IN THE rapidly developing field of cancer genetics, family cancer clinics have evolved to respond to the needs of individuals and families at risk of breast cancer and other common cancers. Risk assessment and genetic counselling are an important focus of these services, and evaluation studies are starting to describe the impact of genetic counselling in terms of improved accuracy of risk knowledge. A small number of studies have also highlighted the needs and expectations of counsellees attending such clinics, indicating the wide range of areas to be covered and perceptions to be addressed [1–4]. For instance, the need for written information before [1] and after [2] the first visit to a familial breast/ovarian clinic has been highlighted, so that women can better question and absorb the information provided.

To date, no publication has described the specific needs or expectations of a cohort of women who are already affected with breast or breast/ovarian cancer, and the paper by Julian-Reynier and associates (insert pages MS 97681 [5]) makes an important contribution to our knowledge. Their questionnaire assessment of affected women carried out in family cancer clinics in six French cities revealed high expectations for prevention options and unrealistic presumptions about the availability of genetic testing. The need for such patients to search for causes of their disease and to understand better their own illness was pointed out by the authors, who stressed that medical explanations may differ from lay representation of the disease. Given that affected women are also the inevitable messengers of information to other family members, the method of communication of cancer risk information is a very necessary focus for future research.

In contrast to genetic counselling for other inherited conditions, breast cancer poses a number of problems because of uncertainties and limitations in risk management. The options for prevention (e.g. chemoprevention or prophylactic bilateral mastectomy) are restricted and unproved. Moreover, mammographic screening in 'at risk' women under 50 years of age has not been evaluated, adding to anxiety about cancer detection. Diagnostic genetic testing for *BRCA1* and *BRCA2* is not yet widely available and other genes are likely to be implicated. Even confirmed gene carriers are left with some uncertainty because of incomplete penetrance of the breast cancer predisposing genes. Thus, risk assessment does not automatically lead to effective strategies for confirmation of risk or further intervention.

Currently variability exists in the provision of surveillance and clinical screening, which, taken with the issues raised above, challenge the value of risk counselling. In response, it is argued that the provision of information is important for psychological well being as well as for appropriate decision making. However, we need to ask certain questions in relation to cancer family services, namely, 'How can attendees most benefit from these consultations?,' 'How should information be communicated?', and 'What are the appropriate outcome measures for evaluation?' These enquiries need to include women at risk who are cancer patients as well as those who are healthy.

There is also an important role for these services to provide a framework for other research, to determine the epidemiology of cancer genes, the biological characteristics of the cancers, the value of screening programmes, the efficacy of drug prevention strategies and the psychosocial costs and benefits of all levels of information giving and intervention. Julian-Reynier and colleagues alert us to the needs of a significant user group with their insights from patients, but they also remind us of our responsibility to provide the best standards of medical management and up to date information [5], which are achieved through these broader research initiatives.

PERCEIVED RISK: IS IT A USEFUL CONCEPT?

Given the medical uncertainties concerning breast cancer risk management, it is indeed valid to research the needs and expectations of attendees, so that misperceptions can be corrected and educational programmes initiated. One common misconception concerns the level of breast cancer risk, both for the general population and for the individual in a cancer-prone family. This is surprising given the amount of publicity about breast cancer, but less than 10% of first time attendees have an accurate estimate of either of these risks before genetic counselling [6]. Research has, therefore, focused on the accuracy of risk perceptions as an outcome of studies evaluating risk counselling, with significant improvements reported in some studies [1, 7], but not others [8]. Given the limited preventive options available to high risk women, is the concept of risk as important as we first thought? In traditional

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genetic counselling, improved accuracy of risk perception may be reflected in subsequent decision making (e.g. family planning), but this outcome is inappropriate in the field of breast cancer.

In an overview of risk concepts, the Dutch psychologist Vlek [9] concludes that 'perceived risk does not seem to be based primarily on statistical considerations, rather it is a multidimensional concept that may be differently weighted by different individuals'. It depends not only on objective information, but on how that information is appraised and processed in light of the context of that individual's life. Losing a mother in childhood, seeing relatives die from breast cancer, approaching the age of a mother or sister when she was diagnosed, may all influence a woman's perceived vulnerability more than a probability estimate. The information becomes 'personally transformed' [10] and accounts for the fact that some women may perceive a low risk as high, while others express relief that cancer is not inevitable, even after being informed of a high risk.

There is evidence from mainstream genetics supporting a theoretical basis to the understanding of variability in risk perception. Shiloh and Saxe [11] identified factors, such as prior expectations, that correspond to categories of the Health Belief Model [12], lending support to the importance of cognitive and motivational factors in accounting for the variance in risk perception in genetic counselling.

It is not surprising, therefore, that individuals may overestimate their susceptibility to cancer because of their past experiences of the disease in the family, or underestimate their risk, perhaps as a means of coping. The risk of cancer may be based more on a 'binary' perception of the risk (either I have the gene or I do not) rather than on a value that takes into account the estimated genetic loading in the family and the degree of penetrance of the genes.

We should, therefore, consider the risks and benefits to those who either underestimate or overestimate a personal risk when considering risk counselling as an intervention. With respect to overestimators, a more accurate risk appraisal may reduce excessive breast checking, prevent an inappropriate demand for genetic testing or preventive surgery and promote more appropriate health care behaviour and surveillance [13]. It has been suggested that such women may also be a valid target for psychological intervention, as they may use a 'monitoring' coping style, making them vulnerable to psychological distress. For women who underestimate, fears that providing accurate risk knowledge would induce anxiety have not been borne out by research in the U.K. [7, 14], so that such estimates may not reflect a necessary coping defence. Thus, it is hoped that the provision of accurate information to this subgroup may lead to appropriate 'breast awareness' and risk management.

These results are encouraging, but affected women have not been systematically evaluated in these psychosocial studies and concern has been expressed about the additional burden of a genetic risk to the breast cancer patient and her family. It is also concerning to note that more specific indicators of cancer worry may not be relieved by genetic counselling [7], irrespective of risk perception, and more research is needed to determine ways of alleviating this.

In considering medical interventions, an understanding of the available information on hereditary breast cancer is more relevant than counsellees' abilities to affirm a statistical probability.

THE PURPOSE OF RISK COUNSELLING

Breast cancer genetic counselling is clearly different from reproductive genetic counselling because of the level of anxiety and threat associated with the disease. Informative results from genetic testing are obtained for only a minority of those who present for risk assessment: the majority of consultees are given only statistical probabilities leaving a degree of uncertainty. Moreover, unlike many inherited disorders, the risk of cancer can never be zero because of sporadic disease.

How can this risk information be used by an individual? Firstly, a programme of surveillance or clinical screening may be offered or recommended and there is no doubt that many women with a family history of breast or breast/ovarian cancer are highly motivated to access such services.

Women's beliefs about other cancer risk factors are highlighted by Julian-Reynier and colleagues [5] and include personal habits such as diet, smoking and alcohol intake. Genetic counselling can help educate women about appropriate risk factors, but there is as yet little evidence that health care behaviour is influenced by genetic consultations. Whilst being 'at risk' may ideally promote adherence to screening programmes, the results of studies in this area have been contradictory and high levels of anxiety about cancer risk may be inhibitory. In the study by Julian-Reynier and associates, stress and the environment were as likely to be considered important risk factors for breast cancer after genetic counselling as before [5]. Clinicians themselves are not in agreement over the importance of certain elements in risk calculation, such as hormonal factors and evidence for others, such as the role of stress, is at best speculative.

Preventive options are even more controversial, with a lack of proven benefit often cited as the basis for not offering prophylactic surgery. Yet the demand for this is gradually increasing and it is essential to verify a woman's cancer history before she seeks out these approaches.

Despite some limitations of risk counselling, the demand for referral to cancer family clinics continues to increase. When faced with the possibility of breast cancer, many women want information to help them manage the risk and some want to be proactive in trying to ensure their future health. Having access to specialist health professionals who understand their needs and concerns can be an important factor in helping these women cope.

COUNSELLEES' INFORMATION NEEDS

Published data suggest that women with a family history of cancer have a wide range of reasons for attending a cancer family service, but their expectations may not be quite in synchrony with those providing the service. For example, our own studies [3, 14] have indicated that up to a quarter of first time attendees specified a wish to reduce their risk of breast cancer as a reason for attending. An equivalent proportion wanted to reduce their anxiety about breast cancer and many women were primarily seeking some form of reassurance, either through mammography, clinical breast examination or from a discussion of their risk. The number of women who indicated that they wanted to be given a personal risk value was extremely high. Interestingly, Julian-Reynier and associates found that less than half their affected sample mentioned this, suggesting different needs in this group [4, 5]. They draw attention to the fact that women with cancer, who form half of all consultees in France, attend mainly for their offspring's sake, whereas healthy clients attend for their own

sake. The implications for communication strategies are a valid target for research.

COMMUNICATION ISSUES

Given the areas in which women 'at risk' want information, discussion and reassurance, and the complexity of risk information to be presented, cancer genetic counselling demands a high level of sophistication in communication skills. Add to this the fact that discussing the family history can be upsetting for consultees, especially if past bereavements are unresolved, the genetic counsellor may require specific skills to handle emotional distress and explore concerns raised. The consultation is not simply one of information giving, yet most attention has been focused on the need for reliable family pedigrees and epidemiological data on which to standardise risk calculation: there are no published data concerning the communication process or the training needs of staff who undertake this.

Data from communication studies in oncology reveal that doctors and nurses find breaking bad news difficult and often fail to detect distress in their patients, so that training initiatives are becoming established to address these problems. In drawing our attention to patients with cancer who attend family cancer clinics, Julian-Reynier and colleagues allude to the sensitivity required in discussion, not least in dealing with the disappointment engendered in some patients by the current lack of preventive strategies for their relatives [5]. We are also reminded that the patients themselves will be the bearers of this genetic information to their families, so that it behoves clinicians to achieve high standards of communication skills in order that the information is imparted in an optimal way.

Finally, Julian-Reynier and colleagues [5] indicate that patients expect and require information about the further management of their cancer risk, given the increased chance of second cancers, so that cancer family clinics must have access to a clinician with the expertise to understand and answer questions concerning oncological management as well as genetic issues. The organisation of future services will need to take into account the varied skills required to meet these wide-ranging needs. It is likely that a number of models could be appropriate, and evaluation will be required to tease out the strengths and weaknesses of different approaches.

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