Communicating Familial Risks: Individual and Community Issues Involved in Cancer Genetics

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A small percentage of all the occurrences of the ‘serial killer’ diseases with which our present-day societies are beset, such as cancer, cardiovascular diseases, infectious diseases and neurological disorders [1], among others, can now be said to come within the scope of predictive genetics. One of the main challenges faced in this first decade of the 21st century is how to integrate the latest genetic knowledge into common medical practice. The introduction of genetic factors into our overall picture of common diseases has greatly enlarged the scale on which traditional genetics work because of the large numbers of people involved in our populations. More than half of all adults in their sixties have at least one case of cancer to report among their first-degree relatives [2], and everybody is likely to be involved in genetic issues if we also include cardiovascular and neurological disorders.

The ability to communicate information about familial risks is therefore a key point worth considering. The papers in this special issue of *Community Genetics* deal with some of the problems involved in communicating breast/colorectal cancer risk information. The authors of these studies all belong to a collaborative network set up within the framework of a European project focusing on cancer risk communication [3, 4].

Introducing new genetic findings into clinical practice by organising specific consultations to inform people

about the cancer risks possibly running in their family is a newly emerging form of clinical practice. It requires the participation of networks of two particular kinds. Those of the first kind are networks of health care providers, namely medical specialists in the fields of cancer and genetics. The first paper in this special issue describes the case of the ‘CRISCOM’ network and explains how seven centres (in Haifa, Hannover, Leiden, Leuven, Marseilles, Manchester and Milan) have been carrying out their clinical and biological practice since the end of the 1990s in Europe and Israel [5]. The authors point out the differences and similarities between countries in terms of the health professionals involved and the laboratory issues arising, as well as stressing the importance of a multidisciplinary approach to the organisation of these activities. This article shows in particular the links existing between research and clinical practice. The second type of network in question is the referring network. The referring providers can be specialists, general practitioners or nurses, depending on how the health care system is organised. Welkenhuysen and Evers-Kiebooms [6] describe here the reactions of general practitioners, nurses and midwives to breast cancer genetic information and, in particular, their attitudes towards the testing and management of high-risk women.

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Cancer genetic consultations are a fairly recent development, but they are still being run on traditional genetics lines in terms of information giving and counselling [7]. How risks should be presented is a complex issue, and Hopwood et al. [8] report here on their clinical experience, presenting risks in terms of proportions (odds) and odds ratios. These authors state that this format is well understood in the United Kingdom. The effectiveness of the various ways of formatting risks is still being discussed in the literature, and women's preferences in various countries have not yet been properly documented [4]. Investigating these preferences will be one of the next steps in the CRISCOM project.

Cypowyj et al. [9] look at the information sources that women use on their own during the genetic testing process. They also describe the interactions which occur between these women and their social network, relatives and doctors when they are exchanging opinions about breast/ovarian cancer genetic testing. Here, the central role of cancer geneticists is highlighted, both as information providers and as the main persons with whom women can discuss what they feel about genetic testing.

The reasons why it is necessary to assess and communicate information about breast/ovarian cancer risks include the fact that this procedure helps to guide the decisions which have to be made about the surveillance and prevention options available. However, recommendations about surveillance and prevention tend to differ from one country to another, even when they are based on similar clinical findings [10, 11]. In this issue, van Dooren et al. [12] present results obtained regarding the link between the frequency with which breast self-examination is carried out by women with breast cancer in their families and their level of anxiety, stratifying the results according to the level of cancer risk. Apart from the fact that women can be guided differently in one country compared to another, or depending on which providers they consult, the finding that high-risk women with high anxiety levels carried out breast self-examination much too frequently is of clinical significance, since it means that highly distressed women should be identified in order to provide them with some extra support. It also suggests that breast self-examination might have stressful side effects.

The psychological and behavioural effects of BRCA genetic testing is a crucial issue on which the results of ongoing prospective studies will throw light. Women's social and preventive behaviour and their perception of the results of these tests as a stressful life event were not found to differ depending on their carrier versus non-carrier status [13].

Since identifying a deleterious BRCA mutation in newly consulting families where a direct gene mutation search is not indicated is an expensive and technically difficult step, the question as to whether this information should be conveyed to the families via the first individual tested is of great importance. Nippert and Schlegelberger [14] have contributed to this special issue by outlining the German approach to cancer genetic testing and describing how some of the individuals involved reacted to the idea of conveying this information to their family. In particular, these authors established that women with positive test results were more reluctant to transmit this information than those with negative results.

After Huntington's disease, which was the precursor, and neurological disorders, which followed on, the field of clinical oncology is one of the pioneer examples of how predictive genetics can be applied to common disorders. The difference here is that the predictive approach makes it possible in the case of familial cancer to diagnose the disease at an earlier stage, with a better prognosis, or even to prevent its occurrence altogether. However, given the undesirable side effects of preventive surgery [15], which is the only strategy reported so far in the literature to be really effective, and the lack of evidence supporting the benefits of the screening strategies available, a great deal of research is still required at both the clinical and community levels. One of the main aims of the present issue is to encourage research workers focusing on these important issues to continue to pursue their efforts along these lines in order to improve our knowledge of the factors involved in the use of these predictive strategies and the advantages to be gained by patients.

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