Genetic testing and breast cancer: the women’s point of view

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Breast cancer is the most common cancer among women in Western countries (130 000 cases per year in Europe) and accounts for 20–25% of all malignancies in European women. In the past few years medical journals have focused greater attention on the quality and quantity of information provided to consumers; there is a general consensus amongst physicians on the importance of having better informed consumers. This change in attitude is influencing greatly the physician–patient relationship and political decisions. Breast cancer associations, like the National Breast Cancer Coalition in the USA or EUROPA DONNA, the European Breast Cancer Coalition in Europe, have pushed for involvement in the discussion of any phase of illness, and have a particular interest in preventive medicine. The identification of high-risk women by genetic testing for BRCA1 and BRCA2 mutations is largely debated, in particular regarding patient counseling, and psychosocial and legislative support. This article reports the different points of view raised by women’s movements, so that useful suggestions may be provided to improve breast cancer prevention modalities.

Key words: breast cancer, counseling, familiarity, women

Introduction
Breast cancer is one of the most important health problems for women in industrialized countries [1]. According to statistics from the World Health Organization’s International Agency for Research on Cancer, more than 216 000 women were suffering from breast cancer in the European Union in 2000, and more than 79 000 women died of this disease. Despite the fact that breast cancer treatments are now less invasive and the survival rate has improved, the impact of breast cancer is still devastating for most women, as well as a burden for other members of the family [2]. All these facts account for the increasing interest surrounding any kind of prevention or early detection methods, and explain why breast cancer must not be considered only a relevant medical problem, but also a human and social issue.

Since a percentage ranging from 2% to 5% of breast cancer (and ~10% of ovarian cancer) can be attributed to mutations in the BRCA1 and BRCA2 genes, genetic testing—a process by which genetic modifications can be sought that are associated with an increased risk of breast cancer—may reveal whether the risk for breast cancer has been passed on through family genes. Although the proportion of breast cancer cases associated with gene mutations can be considered to be low, there is great interest in this issue among women that have experienced breast cancer, and in particular among women with a positive family history of breast cancer [3].

Consumer associations play a relevant role in promoting information and participating in the public debate on health. Most breast cancer associations regularly publish brochures, newsletters or booklets for consumers or patients, and a number of associations have set up call centers to cater for the increased need for information on disease prevention and treatment [4]. This article intends to present and discuss some of the key points of the debate going on within the Italian Forum of EUROPA DONNA, the European Breast Cancer Coalition, with special emphasis on issues relating to genetic testing for breast cancer.

The Italian Forum of EUROPA DONNA
EUROPA DONNA is a non-profit-making organization working autonomously within the European framework. It is a European women’s movement against breast cancer that provides a focus for the exchange of information and experience between members, and serves as a motivating force for combined actions. It is organized as a coalition of individual women and associations involved in the fight against breast cancer, and it is active in 32 European countries [5]. EUROPA DONNA represents the concerns and interests of European women, with particular attention to the need for improvement in or introduction of essential programmes and services in the fight against breast cancer.

The Italian Forum began its activities in Bologna in 1996, when a group of representatives from breast cancer associations, which had already been involved in research programs [6], came together and elected the first National Committee.
The conviction was that associations sharing the same objectives would be stronger if united in a coalition, and that they could more easily achieve results than if they were working individually.

At the moment, the Italian Forum represents the voice of more than 100 breast cancer associations; the coalition does not purport to replace the precious work that these associations carry out locally, namely dealing with all phases of breast cancer, from early diagnosis to surgery, rehabilitation, up to psychological recovery. The results of two surveys regarding the geographical distribution, characteristics, services offered and volume and nature of phone requests for information, document the ever-growing role that Italian breast cancer associations have in the health debate [4, 7].

The main areas of interest of the Forum are increasing public awareness, information and training, and lobbying. The Italian Forum of EUROPA DONNA started its public awareness activities by promoting a Manifesto signed by over 100 famous women. Every year the Italian Forum organizes press conferences, public meetings and television videos on the early diagnosis of breast cancer. Every 2 years since 1999, the Italian Forum has organized a literary award called ‘Before and After’, presented for the best poem and prose writing dealing with the inherent problems of breast cancer as personally experienced by women and the problems faced by their loved ones. All the material collected is a testimony to the different feelings experienced, from rage about the difficulties, to hope for the future, or memories.

Since 1998 the Italian Forum has organized training courses for its activist members on the methodology of clinical research and screening programs (1998 Milan, 1998 Rimini, 1999 and 2002 Naples), which were attended by over 100 activists. The Forum also holds a yearly conference for members of the coalition, with various sections on up-to-date scientific topics. Several brochures are written, printed and distributed by the Italian Forum through the breast cancer associations. Recently, in collaboration with main medical organizations, a brochure called ‘Ten Questions on Mammography’ was published to provide women the opportunity to receive the correct information on this examination modality.

At the beginning of 1997, the results of a survey on the difficulties women meet with in obtaining external breast prostheses were presented during a meeting organized by the Italian Senate, and at the end of that year a new law on the matter was passed. This was the first success of the lobbying activities fostered by the Italian Forum. Lobbying, through the support of all female members of the Italian Parliament notwithstanding their political differences, is among the Italian Forum’s most important activities. In fact, the involvement of institutions both locally and nationally is a means through which results can be achieved to benefit a large number of women. To date, in addition to the revision of the law concerning the provision of external prostheses after mastectomy, the Forum has pressured to change the DRGs regarding breast reconstruction after surgery, and has obtained financial support for radiotherapy and free mammography for all women aged 45–69 years. Owing to the huge discrepancies in terms of assistance and facilities between the various parts of our country, Regional Reference Groups have recently been set up to concert and coordinate the actions of the breast cancer associations belonging to the Italian Forum and politicians at regional level, in order to promote and develop initiatives for better and more uniform care and services for women with breast cancer.

Genetic testing: women’s concerns

One of the assertions of EUROPA DONNA is that completeness, comprehensiveness and correctness of information are a primary and fundamental requisite for knowledgeable involvement in any clinical decision. Accordingly, in a scenario where genetic testing is performed, women should not only receive information that is as precise as possible regarding the value of the test itself in terms of accuracy, but also exhaustive information on the existence and outcomes of possible interventions in case their test is ‘positive’. Moreover, emphasis is laid on the fact that a positive result does not indicate if or when breast cancer will develop, but only documents an altered (i.e. increased) risk of developing the disease. In this and similar situations, the role of breast cancer associations is to ensure that the quality of the information provided be safeguarded by pushing for the use of evidence-based medicine and of simple and clear language [8].

In the case of preventive interventions, i.e. actions aimed at preventing the untimely occurrence of the six ‘Ds’: death, disease, disability, discomfort, dissatisfaction and destitution [9], quality of information is very important in order to reach truly informed consent. As shown recently by Domenighetti et al. [10], the information that women receive on mammography screening is still very unsatisfactory; in spite of the implementation of several breast cancer screening programs and the extensive amount of information conveyed by the media, oncologists, women’s associations and the National Health Service. Most of the American, English, Italian and Swiss women of the sample interviewed overestimated the benefits that can be expected from mammography. In the case of genetic testing, informed consent must be obtained through accurate pre-test counseling, where several items have to be considered. In addition to ad hoc information on the BRCA1 and BRCA2 tests and the implications of the results obtained, women require particular attention on psychological implications, discrimination, confidentiality and sharing genetic test results with at-risk relatives [11].

In general, before taking a genetic test, emphasis should be placed on the fact that a positive result means making important decisions for the future. Several options are usually possible once results are positive, each having a significant burden both from the physical and psychological point of view, since the various options available entail different impacts on the quality of life of the women.

One of the less invasive interventions is a simple ‘surveillance’, with intensive monitoring through clinical and imaging
examinations. Although this kind of intervention cannot be considered to be a truly invasive therapy, it can seriously heighten the stress and anxiety of the individuals involved.

‘Pharmacological prophylaxis’ can also be considered as an option. Unfortunately, at the moment, there is only little evidence regarding its efficacy, while there remain some important risks associated with its use. Hence the decision to start pharmacological prophylaxis should be taken case by case; a woman and her doctor must carefully weigh up the benefits and risks of therapy by considering the case at hand.

The most invasive alternative is ‘prophylactic mastectomy’, which involves an invasive surgical treatment, the efficacy of which is still under discussion [12].

Finally, the last possibility to be taken into account when uncertainty prevails is to enrol in a research study, the aim of which is to test the likelihood of reducing the risk of breast cancer by evaluating the efficacy of alternative options.

In any case, whatever a woman may decide, breast cancer associations have stressed the need to have a well-structured collaboration in place among family doctors, medical specialists (oncologists and surgeons), medical geneticists, psychologists and basic researchers to provide qualified support during and after the decision. Creation of a multidisciplinary team is important to ensure high-quality service. The development of guidelines based on validated scientific results, together with appropriate education and training for health care providers who offer genetic testing for cancer, are advocated to ensure standard treatment, including the access to preventive measures and surveillance options, and equity in the provision of interventions.

Finally, one of the topical issues relating to the results of genetic testing is confidentiality, so that stigmatization of women with a possible diagnosis of breast cancer is avoided. The women of the Italian Forum strongly identify with the position of the American National Breast Cancer Coalition [13]: “To protect women and their families from genetic discrimination based on this newly discovered genetic information, we need powerful laws at both the federal and state levels. Laws covering insurance companies and employers are very complex. To ensure that all women and their families are protected, we need laws prohibiting discrimination by health insurers and employers based on genetic information.” Recently, the ethical aspects of genetic testing have been discussed by the European Group on Ethics in Science and New Technologies of the European Commission, and a document has been published on the discriminatory use of genetic test results [14].

The proliferation of genetic tests calls for effective methods by which to distinguish tests that promote health and prevent disease from those that offer limited benefits or may even be harmful for the general public. The breast cancer associations working with the Italian Forum of EUROPA DONNA have an unremitting interest in the debate on BRCA testing, and are ready to work with physicians, researchers, geneticists, psychologists, nurses and all the other professionals involved so that recommendations and statements may be jointly drawn up to cater more effectively to the needs of women.

References