Friday, 2 October 1998 **08:30-09:30**

DEBATE

Genetic testing: should it be discouraged?

395 INVITED

Contra: Genetic testing should *not* be discouraged

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Purpose: To study the clinical genetic, oncological and psychosocial implications of recent breakthroughs in the field of molecular genetics, in particular the identification of the Breast Cancer Genes BRCA1 and 2.

Methods: Evaluation of our own results in a series of over 450 families with hereditary breast (-ovarian) cancer syndrome (HBC and HBOC) in addition to those reported in the literature.

Results: Recent studies showed that a great majority (75–96%) of first-degree relatives of patients with breast, ovarian or prostate cancer are interested in obtaining information about genetic testing; only 1–5% had no real interest. After detection of a specific gene alteration in a particular family (meaning 60–90% risk of cancer) two thirds of women at risk appeared to opt for presymptomatic DNA-testing already shortly after the DNA-diagnosis. Individuals rated the benefits of testing as significantly more important than the limitations. From an oncological point of view regular surveillance of women at risk from the age of 25 years can result in early diagnosis of breast cancer, which may improve final clinical outcome. In view of a high incidence (40–60%) of ipsilateral and contralateral recurrences/new primary tumors in BRCA1/2 mutation carriers, genetic testing can influence the choice of type of surgery. With respect to prevention prophylactic surgery (frequently showing already premalignant lesions and sometimes even invasive cancers in removed breasts and ovaries) dramatically reduces the risk of cancer, decreases anxiety and prolongs survival. In addition, reduction of risk by tamoxifen recently appeared also to be possible.

Conclusion: Persons at risk of cancer have the right to be informed about risk assessment, surveillance and prevention. Genetic testing can help to make adequate decisions about lifestyle and treatment options saving lives.

396 INVITED

Pro: Genetic testing should be discouraged

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At the identification of three of the breast cancer predisposition genes (BRCA1, BRCA 11, P53) opens up many exciting avenues for research into the molecular mechanisms of the genesis of breast cancer which may ultimately lead to a molecular solution to the problem. Unfortunately there has been so much hype around these discoveries that the lay public have been mislead into believing that genetic testing might be good for them. If a genetic test could be 100% sensitive and 100% specific and if there was an intervention that could lead to the prevention of breast cancer amongst those with the inherited predisposition gene then perhaps genetic testing might be of value to individual members of the public. Unfortunately a negative test in a family with a high prevalence of breast cancer cannot guarantee the absence of genetic predisposition and a positive test for a gene with uncertain penetrance does not guarantee that the woman will develop the disease. Furthermore even those patients with a predictable mutation such as women of Ashkanasi stock cannot assume that genetic testing can have any pay off because as yet we have no guaranteed ways of intervening. Chemoprevention is as yet unproven. Mammographic screening may offer false reassurance and is unlikely to reduce mortality from breast cancer in this young age group and bilateral mastectomy is a cruel intervention which in itself cannot guarantee protection from breast cancer because of residual islands of breast tissue in ectopic sites. Furthermore even submitting oneself for genetic testing may carry a penalty for life insurance and health insurance. Lay women with a familial predisposition to breast cancer should recognise that the subject is in its infancy and that genetic testing must be considered a research enterprise linked in with appropriate research trials on interventions. Marketing kits for the commercial exploitation of these tests should be judged unethical and the only battle to patent the genes for commercial exploitation is a stain on the reputation of biotechnology corporations.