

GENETIC TESTING FOR BREAST AND OVARIAN CANCER

If you have a personal or a family history of breast or ovarian cancer, you may have heard news in recent years about developments in genetic testing. Genetic testing for breast and ovarian cancer is a way to determine if you or a family member carries a gene change (mutation) that increases the risk of these diseases. Genetic testing provides information on the likelihood of developing these diseases in the future. Knowing your cancer risk may help you and your doctor to make better-informed decisions about your health care.

Genetic testing raises many important issues for women and their families. The information below is presented to help you think about these issues. An accompanying leaflet gives information about cancer in general and about breast cancer in particular.

CAN cancer susceptibility be inherited?

In some cases, cancer susceptibility can be inherited. We can attribute approximately 10% of breast and ovarian cancer to single gene alterations that are inherited. That means that 90% of all cases of breast and ovarian cancer are **not** due to an inherited susceptibility from a single gene. The development of cancer is considered a multi-step process. Several different changes have to occur to the DNA before cancer starts. Sometimes, an individual can inherit (from genes passed by their mother or father) one "step" in this process. This makes them further along in the process towards cancer, therefore fewer events have to happen before they develop cancer. They don't inherit cancer itself, but inherit a higher risk of developing cancer than the general population.

HOW are families with hereditary cancer different?

Many of us have family members who have cancer. However, families with inherited cancer susceptibility often have several members with the same type of cancer over several generations. Additionally, there are typically first-degree relatives (mother, sister) who have cancer. Ovarian cancer in two closely related women, because it is uncommon, may more likely be due to inherited susceptibility than by chance. The members of a family who have inherited a gene that predisposes them to cancer usually have cancer diagnosed at an earlier age than expected, and are often affected bilaterally (in both breasts). The genes involved with breast and ovarian cancer are not gender-dependent, so the family history of cancer can appear on the mother's side or the father's side. This means a susceptibility to cancer can be inherited from one's mother or father.

WHAT is the significance of the discoveries of "Breast Cancer genes"?

BRCA1 and BRCA2 are the two major genes responsible for inherited predisposition to breast cancer (and, especially for BRCA1, to ovarian cancer as well). Most women who carry mutations in BRCA1 or BRCA2 have a risk of breast cancer of approximately 80% by age 70, in contrast to the lifetime risk of approximately 8% for most women. In addition, individuals with mutations in BRCA1 carry a risk of ovarian cancer of approximately 44% by age 70, in contrast to the lifetime risk of approximately 1% for most women. An estimated 7% to 10% of all breast and ovarian cancers are thought to be associated with mutations in BRCA1 and BRCA2.

NB: These risk percentages were calculated from high risk family linkage studies, and actual risks may vary.

HOW is testing arranged

Women who have breast cancer, or who are concerned about their risk of developing breast cancer, can seek a referral from their family doctor or specialist to the National Centre for Medical Genetics or to one of our clinics in Cork, Galway or Limerick

Patients referred to one of these clinics will be asked to complete a questionnaire before coming to clinic. The form will ask about your own and your family's medical history, and about your own perception of your risk of developing breast or ovarian cancer. The form will be evaluated by the doctor, who will decide whether you need to be seen at the clinic, or can have your concerns addressed by letter.

Patients seen at the clinic will have a more detailed personal medical and family medical history taken, and will be advised of their risk of developing breast or ovarian cancer. If this risk is high, we may wish to obtain a blood sample from a member of the family who has cancer, to look for changes in the BRCA1&2 genes.

We cannot offer family genetic testing unless we find such a change in your family.

Full counselling will be provided before, during and after all testing. You are free to withdraw from the process at any stage, and this will not affect your medical care.

If a change is found in the BRCA1 or BRCA2 gene, we can offer testing for this mutation to at-risk members of the family.

Knowing your cancer risk may help you and your doctor to make better-informed decisions about your health care. All testing is entirely voluntary, and counselling about the test will be given before any sample is taken, and when the results come back.

Genetic testing raises many important issues for women and their families. This information is presented to help you think about these issues, so you and your doctors can work together to decide if testing makes sense for you.

If you have queries about genetic testing or cancer, please ask the person who gave you this leaflet, or contact the National Centre for Medical Genetics by one of the means listed below.

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