Features

Chroniques

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Popularity of breast cancer program leads to referral protocols in BC

Heather Kent

In brief

A BRITISH COLUMBIA PROGRAM that tests patients for susceptibility to hereditary breast cancer is proving popular, but Heather Kent reports that the testing raises some ethical issues for physicians.

En bref

UN PROGRAMME DE LA COLOMBIE-BRITANNIQUE dans le cadre duquel on soumet des patients à des tests de dépistage de la vulnérabilité au cancer du sein héréditaire se révèle populaire, mais Heather Kent signale que les tests posent des problèmes d'éthique pour les médecins.

A 2-year-old British Columbia program for patients worried about hereditary breast cancer already has a 1-year waiting list. The BC Cancer Agency launched its Hereditary Cancer Program after the *BRCA1* and *BRCA2* genes were identified earlier this decade.

Physicians involved in the program, which began with 3 families in 1996, are now seeing about 15 new families a month from across the province. The program receives no provincial funding — it operates on a 3-year grant from the Canadian Breast Cancer Foundation. Only 126 of the 388 patients referred to the program were seen during a recent 10-month period, creating the 1-year waiting list.

In an attempt to curb the surging numbers, a detailed protocol for referral has been sent to all BC physicians. Included under the new criteria are Ashkenazi Jewish women in whom breast or ovarian cancer has been diagnosed at any age; the breast cancer rate for these women is higher than in the general population. Also included are women under age 36 in whom breast cancer has been diagnosed and women under age 51 in whom ovarian cancer has been diagnosed, as well as men or women under age 51 for whom a diagnosis of colon cancer has been made. Those with strong family histories of cancer — the diagnosis has been made in at least 2 closely related family members — or those with a family history of cancer at a young age are also eligible. (Although admission criteria include cancers other than breast cancer, the program concentrates on the latter because of limited resources and its source of funding. Other cancers are included because the presence of either *BRCA* gene places patients at higher risk for them, too. Dr. Charmaine Kim Sing, the program's medical director, says an estimated 40% to 70% of women with the *BRCA1* gene develop ovarian cancer.)

Patients supplement the referral by completing a detailed screening questionnaire, and 90% subsequently undergo genetic counselling. They are encouraged to bring as many family members as possible to the counselling session, which reviews the family tree and discusses sporadic versus hereditary cancers. Patients learn whether their risk is higher or lower than they thought. A diagnosis of cancer has not been made for most of the women who participate, but they still fear that they may be predisposed to the disease because of their genes, says Karen Panabaker, the program's genetic counsellor.

If genetic testing is considered appropriate for a patient, a family member who already has cancer is the candidate of choice for testing, although 2 people are sometimes tested for increased accuracy. In rare cases DNA from banked blood

Kim Sing (left), McCullum and Panabaker: "The bigger challenge is when you can't find a mutation"

or biopsy tissue is used to study the genes of a relative who died from breast or ovarian cancer. Although the staff have seen women aged from their 20s to their 80s, the "classic scenario" involves a woman in her 30s or 40s who arrives with an older relative who has cancer. With several hundred mutations identified in the *BRCA1* gene and at least 170 for the *BRCA2* gene, analysis of the simple blood test is a complex process. Because of limits on the technology and staff, it can take several months to obtain results.

The absence of a gene mutation presents its own dilemma, says Mary McCullum, the program's nurse educator. "The bigger challenge," she says, "is when you can't find a mutation but the family history suggests that something is probably there."

Staff stress that patients' risk of developing cancer is "not zero" even if genes are normal. On the other hand, discovering a mutation creates a good news/bad news scenario. Although the presence of a *BRCA1* or *BRCA2* gene means a woman is more susceptible to breast cancer, the risk is not absolute. It does, however, mean that the patient is at higher risk of developing other cancers as well.

"The inconclusiveness of the test has to be explained," says Dr. Michael Burgess, a medical ethicist who advises the program team. He says women need to consider their response to a positive result carefully because the medical procedures used to decrease the risk of developing breast cancer are "often more radical than the treatment." Fifty-five patients — 47 women and 8 men — have received results so far, and 33 of the women and 5 of the men had a positive result. Most were "convinced" that they possessed a gene mutation and were "relieved that we had found something," says Panabaker, who, along with Dr. Barbara McGillivray, discloses the results. Panabaker says responses to the news have been "across the board"; seventy-five families are currently awaiting test results.

Kim Sing says men are usually diagnosed with breast cancer at a more advanced stage than women because they don't expect to get the disease. However, because up to 10% of men with a *BRCA2* gene mutation develop breast cancer, the paternal family history is just as important for these patients, she says. (Men with *BRCA1* mutations do not appear to develop breast cancer at an increased rate.)

The consequences that arise when people learn they face an increased risk of breast cancer create a range of ethical dilemmas. As with any kind of genetic testing, the central issue is "how much do you tell and when," says Burgess, who has extensive experience with patients who have Huntington's disease. Whom to tell is also part of that dilemma. Because of concerns about how children would deal with the information, testing is not available before age 18. There are other ethical questions as well. Are patients obliged to disclose their susceptibility to breast cancer to an employer or when applying for disability or life insurance?

Burgess is interested in how the testing affects family relationships, regardless of whether the outcome is positive or negative, as well as whether susceptible patients neglect other components that contribute to breast cancer. He and his students at the University of British Columbia are conducting a study to look into these factors. ?