

Study looks at psychological impact of gene test for breast cancer

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Personal beliefs about inconclusive DNA testing for hereditary breast cancer are associated with cancer-related worry, and such beliefs are an especially strong predictor of whether women had been able to leave the period of DNA-testing behind, reports a study in the October issue of *Genetics in Medicine*, official journal of the American College of Medical Genetics (ACMG). The journal is published by Lippincott Williams & Wilkins, a part of Wolters Kluwer Health.

Published during National Breast Cancer Awareness Month, the study lends new insights into how women cope with the results of BRCA testing for hereditary breast cancer—an increasingly used genetic test in which an "inconclusive" result is common.

Sandra van Dijk, M.A., Ph.D., of Leiden University Medical Center, The Netherlands, assessed psychological adjustment in 215 women who underwent DNA testing for BRCA gene mutations associated with hereditary breast cancer. Of these, 147 women—more than two-thirds of the total—had inconclusive results. The results were positive in 37 women and "true negative" in 31.

Several Factors Affect Adjustment to Uncertainty

An inconclusive result on BRCA testing means the woman carries no genetic mutation currently known to increase breast cancer risk. However, she may still be at increased risk because of unrecognized

mutations, or because of other risk factors associated with familial breast cancer. Of women in the study with inconclusive results, about half were still considered to have more than a 30 percent residual risk of a cancer-causing mutation.

When re-evaluated after four years, none of the three groups had "adverse psychological consequences" of BRCA testing, the researchers write. Not surprisingly, women with negative results had the lowest rates of worry and cancer-related distress. For women with inconclusive results, levels of worry and distress were similar to those who tested positive for a BRCA mutation. In all three groups, the women were less worried than before they were tested.

The study found that the women's beliefs about their inconclusive result had an important impact on their psychological adjustment. "For example, women who report feeling uncertain or ambivalent about their inconclusive DNA test result reported higher levels of worry and distress," Drs. Van Dijk and colleagues write. The women's beliefs about their inconclusive results were "very strongly related" to whether they had come to terms with their result and its implications for breast cancer risk.

The link between beliefs and psychological adjustment remained strong even after accounting for the women's levels of worry and distress. "Cancer-related worries and distress may provide an important but incomplete picture on how women adapt to their inconclusive result," according to the researchers. "[W]omen differ in whether they can cope with the uncertainty of an inconclusive result."

Other important factors included whether the women were actually tested themselves or had received inconclusive results from an affected relative who was tested. Women who were not personally tested were more likely to believe they might have inherited a BRCA mutation,

which in turn was related to lower psychological adjustment.

Implications for Genetic Counseling

As in other genetic tests, the psychological impact of BRCA testing is an important consideration. Previous research has found that BRCA testing does not generally lead to high levels of psychological distress. However, few studies have focused on adjustment in the large number of women with inconclusive results.

Dr. van Dijk and colleagues believe their results highlight the role played by genetic counselors in ensuring that women accurately understand their BRCA test—including the uncertainty associated with inconclusive results. They write, "Not only must women's expectations before BRCA testing be addressed, but also the issue of how women are planning to come to terms with an inconclusive result."

"Over 200,000 women will be diagnosed in the U.S. this year with breast cancer and a significant minority of them will have developed cancer because they harbor a BRCA1 or BRCA2 mutation," comments Dr. James P. Evans, Editor-in-Chief of *Genetics in Medicine*. "Sequencing of these genes has been a boon to patient management and to their family members but such testing comes with a psychological price. In the current issue of GIM, van Dijk et al. explore the psychological effect of discovering such ambiguous and potentially disturbing results."

Source: American College of Medical Genetics

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