BY DEANNA FRANKLIN
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Patients at high risk for breast cancer and ovarian cancer are not getting the full story from the genetic tests now being carried out in the United States, according to a study by Tom Walsh, Ph.D, and his associates.

Genetic testing is recommended for women with family histories of breast and ovarian cancer that suggest they may have inherited BRCA1 or BRCA2 gene mutations. To determine the frequency and type of cancer-predisposing gene mutations that are undetected by the most commonly used genetic tests, Dr. Walsh and his associates enrolled 300 representatives of cancer-afflicted families, or “proband” families harbored 28 different mutations,” the researchers concluded.

Currently, conventional genetic testing for BRCA1 and BRCA2 mutations are done almost exclusively in the United States by one commercial laboratory in Utah. According to the researchers, this lab uses short-range polymerase chain reaction (PCR) followed by genomic sequencing to test for five specific larger mutations in BRCA1. However, recent evidence has shown that many mutations go undetected by PCR, and as a result many high-risk patients are given a negative (wild-type) genetic test result, noted Dr. Walsh of the departments of medicine and genome sciences at the University of Washington, Seattle, and colleagues.

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The lifetime risk of breast cancer may be as high as 80% among U.S. women with BRCA1 and BRCA2 mutations; the lifetime risk of ovarian cancer is greater than 40% for women with BRCA1 mutations, and greater than 20% for those with BRCA2 mutations. Mutations of CHEK2 may double the risk of breast cancer, and—although extremely rare—inherit mutations of TP53 in families with Li-Fraumeni syndrome, and PTEN in families with Cowden syndrome are tied to high risk of early-onset breast cancer.

“As more breast cancer susceptibility genes of different penetrances are identified, clinicians will be increasingly challenged to offer the most appropriate genetic tests, to assist patients in interpreting the results, and to optimize risk reduction strategies,” the researchers concluded.

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