Researcher contributes to debate on breast cancer gene screening

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There has been much recent debate on the benefits and risks of screening for breast cancer using BRCA1 and BRCA2 mutations in the general adult population. With an estimated 235,000 new breast cancer diagnoses each year in the U.S. and more than 40,000 deaths, it is clearly important to be able to determine which women may be genetically predisposed to breast cancer.

Glenn E. Palomaki, PhD, associate director of the Division of Medical Screening and Special Testing in the Department of Pathology and Laboratory Medicine at Women & Infants Hospital of Rhode Island has recently published an invited commentary in the November issue of *Genetics in Medicine*. The commentary is entitled "Is it time for BRCA1/2 mutation screening in the general adult population? Impact of population characteristics."

A family history of breast or ovarian cancer or a personal history of early-onset cancer are strong risk factors for breast cancer. Systematic criteria when caring for a patient with a positive family history have been well established by such agencies as the U.S. Preventive Services Task Force and the National Comprehensive Cancer Network.

Dr. Palomaki said, "With the identification of the tumor suppressor genes BRCA1 and BRCA2 in the 1990s, the scientific community has extensively explored both the personal and population impact of carrying a deleterious mutation in these genes. Any new population-based screening test, such as testing for BRCA1 and BRCA2 mutations, requires consideration of key performance characteristics that evaluate both strengths and shortcomings before its introduction."

In his commentary, Dr. Palomaki cited two recent publications that present perspectives on routine, population-based screening for breast cancer using BRCA1/2 mutations in different populations.

"Together, these two publications offer an unusual opportunity to compare and contrast how distinct population differences, such as the mutations carrier rate, might influence the feasibility of population-based screening," said Dr. Palomaki. "Because founder mutations are more common in Ashkenazi Jewish women, are more easily identified and account for a higher proportion of all breast cancer cases, pilot trials in that population are indicated before launching widespread screening in Israel to identify and resolve implementation issues. Such screening in the United States, however, is more complicated, tilting the balance away from routine population screening, as least for the moment."

Provided by Women & Infants Hospital