Breast cancer patients with high risk gene diagnosed six years earlier than generation before

October 9 2009

Women with a deleterious gene mutation are diagnosed with breast cancer six years earlier than relatives of the previous generation who also had the disease and/or ovarian cancer, according to new research from The University of Texas M. D. Anderson Cancer Center.

The findings, presented in a poster session at the 2009 Breast Cancer Symposium, could have an impact on how women at highest risk for the disease are counseled and even screened in the future, explained Jennifer Litton, M.D., assistant professor in M. D. Anderson's Department of Breast Medical Oncology.

"In our practice, we've noticed that women with a known deleterious BRCA gene mutation are being diagnosed earlier with the disease than their moms or aunts," said Litton, M.D., the study's first author. "With this study, we looked at women who had been both treated and had their BRCA testing at M. D. Anderson to determine if what we were seeing anecdotally was consistent scientifically."

It's estimated that five to 10 percent of all breast cancers are associated with either the BRCA1 or 2 mutation, both of which are associated with an increased risk for breast and ovarian cancers. According to the American Cancer Society (ACS), women with BRCA1 or 2 have a 60 percent lifetime risk of developing breast cancer, compared to a 12 percent risk for women in the general population.

Given their greater risk, women with known BRCA mutations and/or whose mothers and/or aunts from either side of the family have the mutation are screened beginning at age 25. In 2007, as a complement to mammography, ACS guidelines added Magnetic Resonance Imaging (MRI) in the surveillance of these women at highest risk, as MRI is thought to catch smaller tumors even earlier. Consideration of prophylactic mastectomies is also a component of their surveillance, said Litton.

"Currently, BRCA positive women are counseled that they won't need to worry about breast cancer until a certain age. However, our findings show that we may actually start seeing the disease even earlier in future generations. We need to make changes accordingly in order to best advise and care for these women at greatest risk," Litton said.

For the retrospective study, the researchers identified 132 BRCA positive women with breast cancer who participated in a high-risk protocol through M. D. Anderson's Clinical Cancer Genetics Program between 2003 and 2009. Reviewing each woman's pedigree (family tree), 107 were found to have a female family member in the previous generation who also had a BRCA-related cancer, either breast or ovarian. Age at diagnosis, location of mutation and birth year were recorded in both the older (gen1) and younger (gen2) women.

The study found that in gen2, the median age of diagnosis was 42, compared to age 47 in gen1. In comparing generations within a family, the median difference was six years. Comparing decades of birth in all born after 1950, gen2 were diagnosed earlier than gen1. When stratified by BRCA1 or 2 mutation, the median age of diagnosis in BRCA1 and BRCA2 positive gen2 women was 42 and 44, respectively, and 43 and 50, respectively in BRCA1 positive and BRCA2 positive gen1 women.

"These findings are certainly concerning and could have implications on

the screening and genetic counseling of these women," Litton said. "In BRCA positive women with breast cancer, we actually might be seeing true anticipation - the phenotype or cancer coming out earlier per generation. This suggests more than the mutation could be involved, perhaps lifestyle and environmental factors are also coming into play."

The research reconfirms that women with BRCA mutations should continue to be screened per the guidelines - mammography, MRI and consideration of prophylactic surgeries - yet perhaps with increased suspicion and even at an earlier age, said Litton, who notes that the addition of MRI screening may account for some of the change in diagnosis seen in the study.

Further analysis is needed given the relatively small number of women in the cohort and the possibility of recall bias, as the gen2 women were providing their family histories, Litton explained. As follow up study, Litton plans to look into biological basis for potential earlier diagnosis.

Source: University of Texas M. D. Anderson Cancer Center (news : web)

Citation: Breast cancer patients with high risk gene diagnosed six years earlier than generation before (2009, October 9) retrieved 24 April 2024 from https://medicalxpress.com/news/2009-10-breast-cancer-patients-high-gene.html

This document is subject to copyright. Apart from any fair dealing for the purpose of private study or research, no part may be reproduced without the written permission. The content is provided for information purposes only.