Genetic Testing for Breast Cancer Could Benefit Minorities but is Underused

Submitted by hareyan on Mar 16th, 2006
Posted under: Breast Cancer

Brest Cancer Test

Ten years after the identification of the first breast cancer susceptibility genes, so few high-risk minority women have received genetic counseling or testing that the standard methods of calculating risk have not been validated in these groups, and the results of genetic testing can still produce surprises.

In the October 19, 2005, issue of the Journal of the American Medical Association, a research team from the University of Chicago reports for the first time that the predictive model used by genetic counselors to assess risk - based on a family history of breast or ovarian cancer and age of diagnosis - works just as well for families of African ancestry. They also found, however, that the spectrum of mutations that occur in African-Americans is "vastly different."

"Access to genetic counseling and testing are an important part of cancer control," said Olufunmilayo Olopade, MD, professor of medicine and director of the Cancer Risk Clinic at the University of Chicago Hospitals. "But in this fast-moving area of medicine, some ethnic minorities are being left behind. We need to encourage high-risk women from all ethnic groups to get counseling, and we need to learn more about what specific test results mean for each racial or ethnic group."

African-American women are at higher risk for early-onset breast cancer. Many of the tools used to estimate risk, however, such as the widely used BRCAPRO statistical model, were developed based on trials involving primarily women of European descent, especially Ashkenazi Jewish women.

So Olopade, Rita Nanda, MD, and colleagues at the Cancer Risk Clinic at the University of Chicago Hospitals, pulled together 10 years of counseling and testing data from the clinic - supplemented by data from three other U.S. clinics - all of which attract diverse groups of patients.

The researchers studied the woman at greatest risk from 155 families (117 from the University of Chicago Hospitals, plus 38 from Mayo Clinic, Rush University Medical Center and UCSF) who sought genetic testing because they had two or more close relatives with breast or ovarian cancer.

They used this data to determine how well BRCAPRO could predict BRCA1 and BRCA2 mutations, based on family history, among high-risk individuals of both European and African ancestry. They also looked at the range of mutations found in various ethnic groups.

The first thing they noticed was very few minority women sought counseling or were referred for testing. Out of 155 families, three were Hispanic and two were Asian. Although about half of the patients seen at the University of Chicago Hospitals are African-American, less than one-third of the families who sought testing were of African ancestry.

They did confirm that BRCAPRO worked just as well for African-Americans, as it did for other populations. "Irrespective of ancestry," the author said, "early age of diagnosis and a family history of breast and ovarian cancer are the most powerful predictors of mutation status."

Each additional case of breast cancer in a first- or second-degree relative increased the odds of a mutation by 62 percent. Each additional case of ovarian cancer raised the risk by 146 percent. But any increase in the age of a relative's cancer diagnosis reduced the odds by 10 percent.

When they looked at DNA test results, however, they found that the spectrum of mutations was very different for families of African as opposed to European ancestry.

"These mutations are inherited, so they tend to reflect a person's racial and ethnic ancestry," Olopade said. The Ashkenazi Jewish mutations are the most common and consistent, but there are distinctive "founder" mutations in BRCA1 and BRCA2 associated with other European ethnic groups, such as the Germans, Dutch, or Scots.

As expected, damaging mutations in BRCA1 or BRCA2 were most common in Ashkenazi Jewish families.

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Sixty-nine percent of high-risk women tested (20 out of 29) had a mutation, and 85 percent of those mutations were one of three well-documented variants associated with this ethnic group.

Deleterious mutations were also common in non-Jewish, non-Hispanic whites. In this group, 46 percent of women tested (36 out of 78) had a mutation known to be deleterious. Many of these could be traced to "founder" mutations from ethnic backgrounds.

Documented mutations already known to be harmful, however, were less frequent among African-American women. In this group, despite having a strong family history of breast cancer, only 27 percent of those tested (13 out of 43) had a mutation known to increase cancer risk.

African-American women, however, were almost four times as likely as non-Jewish, non-Hispanic whites (44.2 percent versus 11.5 percent) to have other variations in these genes, mutations that have not yet been characterized or linked to disease.

"Some of these variants are probably benign," said Olopade. "And some of them probably contribute to susceptibility."

The common, well-known "protein-truncating" mutations, such as those that afflict Ashkenazi Jewish women, produce a nonfunctional protein with large segments missing. The African-American variants appear to cause smaller changes that may impair, but not shut down, protein function.

The average age of diagnosis for all families in the study was 46. For African-American women with mutations known to be harmful, it was 43.6. For those with "variants of undetermined significance," it was 50.5. For those with no identifiable mutation, it was 42--the youngest of all.

The finding that women without mutations in either BRCA1 or BRCA2 get the disease so early suggests that African-American families with multiple early breast cancers have harmful mutations in another, as yet unidentified, breast cancer susceptibility gene.

"We still have a lot to learn about the ties between genetics and breast cancer in African-Americans as well as many other minority groups," Olopade said. "But we already know enough about the risk factors and the disease to help those most at risk by designing protocols for prevention and early detection."

The take home message, she added, is that women with a family history of breast and/or ovarian cancer should meet with a genetic counselor to have their risk assessed, consider genetic testing, and take appropriate precautions.

The National Cancer Institute, the Falk Medical Research Trust, the Breast Cancer Research Foundation, the Entertainment Industry Fund, National Women's Cancer Research Alliance and the U.S. Department of Defense supported the research.

Source: University of Chicago Hospitals

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