Study reveals young women with the BRCA mutation feel different and misunderstood

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(Medical Xpress)—A study authored by Rebekah Hamilton, RN, PhD, associate dean of the Rush University College of Nursing, found that young women with a BRCA1 and BRCA2 gene mutation felt different and misunderstood.

Women who inherit a BRCA1 mutation have a significantly increased risk of developing breast, ovarian cancer or both. A mutation in BRCA2 increases risk of melanoma and cancers of the pancreas, stomach and gallbladder. While genetic testing may help a woman manage her risk, she may also face complicated decisions regarding relationships, childbearing and a career. The study is published in the October issue of the *American Journal of Nursing* (AJN).

"Although there have been advances in determining a person's genetic risk for disease, little is still known about the psychosocial consequences among young women who carry the BRCA 1 and 2 mutations," said Hamilton. "While being told you have a disease is distressing, learning that you carry a gene mutation that increases your risk of disease as well as that of your siblings and children raises other, more complex, issues."

In 2006, participants with breast cancer and/or BRCA gene mutations were recruited from websites for women. Forty-four women ages 18 to 39 from 22 states and Canada who were found to carry a BRCA mutation were interviewed by phone or e-mail. A qualitative, grounded theory analysis was performed focusing on the participants' being young

and having had genetic testing for the BRCA mutation. The findings focused on three characteristics of the participants—whether or not they were married, had children or had breast cancer and how those characteristics were affected by the women's knowledge of their genetic risk.

Among the 13 unmarried participants, issues included when to tell a new partner about your genetic risk; how early in a relationship to discuss having children or plans for prophylactic mastectomy or oophorectomy. Young women showed concerns surrounding the impact of pregnancy on cancer development, the disruptions on relationships and a sense of discrimination from one's peers. Some expressed a feeling of being less perfect than other family members.

Many of the 24 participants who had children reported "staying alive" for their children as a primary goal and expressed concern and guilt that they might have passed the mutation to their children; the childless women reported an urgency to have children.

Of the 21 who had a breast cancer diagnosis, several said knowledge of their genetic risk influenced their decision to have the unaffected breast removed prophylactically.

"While learning that you have the mutation can empower a woman to reduce her risk of developing cancer or detect it in its earlier stages, it can cause her to worry for herself, her children and other family members," said Maureen Shawn Kennedy, MA, RN, editorial director and interim editor-in-chief of *AJN*. "Nurses need to recognize a patient's knowledge of genetic risk before testing, and consider such factors as woman's age, marital status, breast cancer diagnosis, presence or absence of children, family history of breast cancer, family's response to genetic testing and choices of surveillance as important aspects of care."