



Beyond *BRCA1* and *BRCA2*: Other Gene Mutations Linked to Cancer Risk

Many women with a family history of breast or ovarian cancer choose genetic testing to find out if they have inherited *BRCA1* or *BRCA2* gene mutations, which could put them and their family members at a higher risk for these cancers. Now a panel test – known as BROCA – is available for women to learn if they have other gene mutations linked to breast and ovarian cancer.

The panel includes mutations in about 25 genes associated with an increased risk for breast or ovarian cancer, and that number is steadily growing. Many of them, including *PALB2* and *BARD1*, were identified in Dana-Farber laboratories.

But Judy Garber, MD, MPH, a breast oncologist and director of the Center for Cancer Genetics and Prevention in the Susan F. Smith Center for Women's Cancers at Dana-Farber, advises women and

their physicians to consider this broader genetic testing very carefully. While the information helps assess increased risks of cancer, researchers are still trying to understand exactly how to manage those risks. Unlike *BRCA1* and *BRCA2* mutations, these newly identified gene mutations have not yet been the focus of long-term, large-scale studies that help clarify effective risk reduction strategies.

"With *BRCA1* and *BRCA2* gene mutations, we're not happy we have to recommend surgery so often to prevent cancer, but we know it works," says Dr. Garber. "Now we have newly identified genes and significantly fewer people who've been tested for these genes. We haven't had time yet to do the work that's needed to tell them how much risk is associated with these mutations."

However, Dr. Garber says we can expect progress much faster today

than in previous years. "Many more women are being tested and agreeing to participate in research, and the technology networks for sharing information are much better."

Who should consider this broader testing? It's advisable for women who have had an ovarian or breast cancer diagnosis at any age, and for certain women who have had a cancer diagnosis before age 50, regardless of their family history. It's also recommended for women who have a family history of cancer that is not explained by other genetic testing. Genetic counselors can provide more personalized guidance.

"The goal of cancer genetics is twofold: to help cancer patients who already have cancer, and to prevent cancer in their relatives," Dr. Garber points out. "I'm confident these new technologies will ultimately enhance our ability to do both."