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Study shows families don't understand genetic test results or their implications

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A study done by researchers at Fox Chase Cancer Center shows that many relatives of patients who undergo testing for a gene linked to breast and ovarian cancers misinterpret the results, and less than half of those who could benefit from genetic testing say they plan to get tested themselves—despite the fact that knowing your genetic status may help catch the disease in its earliest stages. The study results will be presented on Thursday, December 12 at the 2013 San Antonio Breast Cancer Symposium.

"People don't always understand genetic information, so there's confusion," says study author Mary B. Daly, MD, PhD, chair of the Department of Clinical Genetics at Fox Chase. "Family members are either not understanding what they're hearing, not realizing it has implications for them, or they're not hearing it at all."

For a long time, Daly says she "naively" assumed that, once one family member knew whether or not they carried genes linked to breast and ovarian cancers—known as BRCA1/2—their entire family would understand the result, and what it meant for their own genetic risk. "Over time, we realized that wasn't happening, or it wasn't happening very well."

Some genetic information is straightforward, says Daly. For example, when a woman learns she carries BRCA1/2 that means her parents, siblings and children may also carry the gene. But there are more "indeterminate" results, which are harder to interpret, she adds. If a

woman with a strong family history of breast and ovarian cancers tests negative for the BRCA1/2 genes, that does not mean her relatives are not at risk, says Daly—her siblings could still carry the gene, or there could be additional genes present that predispose them to cancer that clinicians don't yet know how to test for.

"When you look at some of these families who are so full of breast and ovarian cancer, and the person tests negative, you think there's got to be something going on here. We just can't find it. That's a difficult thing for someone to explain to a relative," says Daly.

To understand better what was (and was not) being communicated after people underwent genetic testing, Daly and her team called 438 relatives of 253 people who had undergone genetic testing and said they'd shared their results. More than one-quarter of family members reported the test result incorrectly. They were most likely to understand positive results—like their family member carries the BRCA1/2 gene. But only 60% understood the so-called "indeterminate" results, where their relative tested negative for the gene but they and other family members could still be at risk. Nearly one-third said they had trouble understanding the result.

Concerningly, only half (52%) of family members whose relative tested positive for the BRCA1/2 gene said they planned to get tested themselves. Among those whose relative tested negative for the BRCA1/2 gene, but knew the gene was present in their families (meaning they could still carry the gene), only 36% said they were going to find out their own genetic risk. "These findings imply the family members did not fully understand the significance of these results for their own risk," says Daly.

People were more likely to share their results with adult children than parents or siblings, and particularly with female relatives. "Over and over

you hear people say 'I'm doing this for my children's sake,'" says Daly.

As part of the study, Daly and her colleagues had asked half of the people getting tested to participate in two coaching sessions to help them communicate their results to relatives, such as through role playing. However, these people were no more likely to communicate the result of their tests than people who had simply sat through educational sessions about overall health. "It didn't matter which group they were in, unfortunately," says Daly. "That disappointed me."

But it also inspired her to develop the next project—exploring the effect of directly reaching out to the relatives of someone who underwent genetic testing (with that person's permission), to see if hearing the results from an expert who's not personally involved in the situation helps family members understand what they mean.

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