Study reveals why almost half of patients opt out of comprehensive cancer testing

May 7 2015

Some at-risk patients opted out of comprehensive cancer gene screening when presented with the opportunity to be tested for the presence of genes linked to various cancers, according to a recent study led by researchers at the Perelman School of Medicine at the University of Pennsylvania and the Basser Center for BRCA in Penn's Abramson Cancer Center. Concern for uncertainty and potential distress were cited among the most common reasons to refuse testing. The results, published in *Genetics in Medicine*, were released just weeks ahead of an announcement of the online availability of low-cost genetic testing for breast and ovarian cancer mutations. Authors say the results suggest that patients have varying interest in "gene panels" when they are informed of the potential risks and benefits, reflecting the current need for pre-test counseling when genetic panel testing is considered for at-risk patient populations.

The study found that of the 49 patient participants, all of whom have a family or personal history that puts them at-risk for development breast and other forms of cancer, more than one-third declined multiplex testing. Multiplex testing allows for the simultaneous analysis of alterations in multiple cancer-related genes, and is an alternative to targeted tests that screen for individual forms of cancer. A targeted test might look for inherited mutations in the BRCA1 and BRCA2 genes, for example, which increase the risk of breast, ovarian, and other types of cancer. Some patients also declined the study all together because they were concerned about the uncertainty or distress with testing.

"Traditional targeted genetic tests, such as those for BRCA1/2 mutations, which test for a small number of similar types of genes, can identify beneficial next steps, such as greater screening frequency, or in some cases having the breasts or ovaries removed," said lead author Angela R. Bradbury, MD, an assistant professor of Hematology/Oncology in Penn's Abramson Cancer Center. "But, we don't yet know enough about many of the genes on these panels, so multiplex testing may cause undue anxiety and stress over findings which may turn out to be benign. In addition, patients may take inappropriate risk-reducing or screening interventions based on very limited information."

In the Penn-led study, the most frequent reasons for declining multiplex testing were concern about information overload and concerns about receiving uncertainty regarding the results or medical recommendations. One current criticism of multiplex testing is that it can identify gene variants that may be precursors to or signs of serious forms of cancer for which effective treatments either do not currently exist or are often radical (such as preventive removal of the stomach). In some cases physicians have few remedies to offer except to counsel patients to wait until more knowledge is gained.

In the new study, some patients elected not to participate in the study because they were not interested in testing that could be associated with uncertainty. All of the 49 patients who enrolled received pre-test counseling, which described the advantages (such as early detection) and disadvantages (including increased worry and uncertainty) of the testing. Following the counseling, 16 participants declined multiplex testing or being told their results. Of the 33 who proceeded with multiplex testing, 16 participants tested negative for any cancer-related variants, five tested positive, and 12 were found to have gene variants of uncertain significance (meaning that it is currently unclear if the changes are benign or tied to cancers).

Overall, researchers found that general anxiety decreased significantly after pretest counseling.

Anxiety, depression, and uncertainty, did not significantly increase after learning multiplex test results.

"One note of caution about our findings is that selection bias may be at work since those who thought that they would become worried or anxious excluded themselves from the testing altogether," said the study's senior author Susan M. Domchek, MD, the Basser Professor of Oncology in Penn's Abramson Cancer Center, and executive director of the Basser Center for BRCA. "What these results show, however, is that many patients who receive pre-test counseling, and are therefore better able to understand and interpret their results, do not experience significantly more distress after testing."

The study results were issued just weeks before an announcement about the online availability of low-cost genetic testing for breast and ovarian cancer mutations. "It is important that at this time, this type of genetic testing for cancer be carried out in concert with a genetics professional," said Bradbury. "Our study is the first of its kind to assess how patients react to these types of tests. Given our finding that patients have variable interest in testing and tolerance for uncertainty, it is best for patients to proceed with testing only after understanding the potential risks and limitations. Ongoing larger studies will help us better understand how people respond to these results, particularly when there is some uncertainty regarding the result or the optimal medical management."

Provided by University of Pennsylvania School of Medicine

patients-opt-comprehensive-cancer.html