A recent study has shown that health behaviors are not significantly altered by the receipt of personal genetic testing indicating a modestly elevated risk of breast, prostate, or colorectal cancer (J Clin Oncol [published online ahead of print December 12, 2016]. doi: 10.1200/JCO.2016.67.1503). The current report is part of the Impact of Personal Genomics (PGen) Study, which is a prospective, longitudinal cohort study examining psychosocial, behavioral, and health outcomes related to direct-to-consumer (DTC) personal genomic testing.

The analysis was performed to determine whether patients who received elevated single-nucleotide polymorphism (SNP)-based cancer risk estimates were more likely to change health-related behaviors compared with those who received an average-risk or low-risk estimate. SNPs are genetic variations that represent a difference in a single nucleotide in the DNA and are present in at least 1% of the population.

“Starting in 2013, DTC health-related genetic testing was suspended by the FDA [US Food and Drug Administration] pending further review of data,” says Stacy Gray, MD, lead author and associate professor in the division of clinical cancer genetics at the City of Hope Cancer Center in Duarte, California. “Therefore, the test analyzed in this current study is not currently available to consumers; however, regulations are evolving with the DTC genetic testing industry and there is the possibility that health-related tests will be available in the United States, like they are in other countries, in the future. Our study adds to the body of literature evaluating effects—potentially adverse or beneficial—of such testing.”

Study Details
New customers from March through July 2012 of 2 genetic testing companies—23andMe and Pathway Genomics—were invited to complete 3 Web-based surveys: at baseline before genetic cancer risk results were received and at 2 weeks and 6 months after receiving results. Surveys included questions regarding diet, exercise, and use of supplements as well as mammography, colonoscopy, and prostate-specific antigen (PSA) testing history. The results of the genetic tests were categorized as having either an elevated or nonelevated risk of breast, prostate, or colorectal cancer. Participants who underwent SNP testing at either company received a single genetic cancer risk estimate based on a genotyping profile of multiple SNPs for breast (women only), prostate (men only), and colorectal cancer. The analysis for this study was performed on 762 patients with completed surveys and demographic data available. Of these, 438 patients had breast cancer risk estimate results, 281 had prostate cancer risk estimate results, and 719 had colorectal cancer risk estimate results available. Elevated risk estimates for breast, colorectal, and prostate cancer were observed in 12%, 24%, and 23% of participants, respectively.

Cecelia Bellcross, PhD, MS, CGC, assistant professor and director of the genetic counseling training program at Emory University School of Medicine in Atlanta, Georgia, who was not involved in this study, says she has concerns regarding the clinical usefulness of SNP testing. “The SNP profile has low predictive value. The SNPs that have been identified account for a minority of the heritable cancer risk. It is like trying to put a puzzle together when you only have 10% of the pieces. Because the risk predictions based on SNP profiles have unclear implications, there is no standard recommendation regarding medical management based on the results.”

At baseline, 68% of participants reported using vitamins or supplements, 43% said they met the dietary recommendations outlined by the

KEY POINTS
- SNP-based cancer risk estimates do not appear to affect health-related behaviors positively or negatively.
- DTC genetic testing will continue to evolve and become more complicated, and the health care provider is obligated to educate patients and refer them to genetic counselors appropriately.
Centers for Disease Control and Prevention (CDC), and 35% stated they met CDC exercise recommendations. Also at baseline, participants aged 50 years and older reported high rates of being up to date with CDC recommendations for cancer screening (mammography in 97%, colonoscopy in 75%, and PSA screening in 80%).

On univariate analysis, the 6-month survey results demonstrated no significant differences between those with high or low cancer risk estimates with regard to changes in diet or exercise habits. Supplement use was found to be significantly increased in participants with a high prostate cancer risk score compared with those with a low risk score among those individuals who reported supplement use at baseline, but this association was not observed for breast or colorectal cancer risk scores.

Furthermore, screening behaviors at 6 months were not found to be significantly different between high-risk and low-risk groups for breast and colorectal cancer, but significantly more men with high prostate cancer risk scores had undergone PSA screening among those who did not report PSA screening at baseline. Not surprisingly, for all 3 cancers, those patients who had undergone screening the year before the survey were most likely to report being up to date with screening at 6 months.

A multivariable logistic regression analysis also was performed that included multiple health-related behaviors and demographic data. Those participants with elevated risk scores were not significantly more likely to change their diet, exercise, or supplement habits, or to engage in advanced care planning or undergo cancer screening. The exception was that those individuals with an elevated prostate cancer risk were more likely to increase supplement use (22% of participants at elevated risk vs 8% of those not at elevated risk [adjusted odds ratio, 3.43]).

Implications
The authors report that although some other studies have shown that patients have reported changes in their health habits after genetic testing, recent reviews and meta-analyses have been in line with the findings of the current study indicating a minimal effect in terms of positive behavior changes or, importantly, of causing any unintended adverse effects, such as unnecessary subsequent medical testing. The high screening rate of participants at baseline may have made it more difficult to observe any post-test differences, they note.

“The main concern motivating this study was that people would see the results and then get tests they did not need,” says Dr. Gray. “However, we did not see evidence of harm. Behavior was not changed in any substantive way in relation to the test results. It does not seem that patients were inappropriately using the test results to undergo unnecessary medical testing.”

Dr. Gray says that, for the most part, the tests in the current study were abnormalities with less well-defined relevance than a high-risk abnormality such as a BRCA mutation. “The medical community does not have clear recommendations for them, which may contribute to the lack of behavioral changes,” she adds.

In addition to the potential for patients to seek extra screening that is not evidence based, the opposite effect could occur from DTC personal genetic testing. “A patient may get a false sense of safety from a negative or low-risk test,” says Dr. Bellcross. “For instance, if a BRCA SNP profile is negative, the patient may feel this means they do not have a BRCA mutation and minimize the importance of a strong family history. They may not have an understanding of the differences in actual gene sequencing and the SNP-based other testing.” Dr. Bellcross says she also is concerned about the possibility of increasing health care disparities because the testing will be performed largely among individuals of higher socioeconomic status.

Genetic counselors in the community setting are actively involved in supporting patients in testing, and can be valuable resources for patients and health care providers. Nancy Cohen, MS, CGC, a genetic counselor at Northern Westchester Hospital in Mt. Kisco, New York, is one such individual.

“It is much more relevant to look at the entire individual with a complete personal and family history to assess hereditary risk than with any testing in isolation,” says Ms. Cohen. “For any genetic test, you have to understand what you are going to do with the results—positive, negative, or uncertain—and what impact that result may have on you and your family,” she says.

Dr. Gray notes that although the particular test studied in the current article presently is not available, DTC genetic testing is expanding with changes in regulations and continues to become more complex, compelling continued research regarding how best to support informed patient decisions concerning DTC personal genomic testing.

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