FOR IMMEDIATE RELEASE
December 13th, 2016
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Studies Probe Value and Impact of Direct-To-Consumer Genetic Testing

A series of scientific reports from the Personal Genomics (PGen) Study reveal insights into patient perceptions and experiences with direct-to-consumer genetic testing

Despite being on the market for nearly a decade, direct-to-consumer (DTC) genetic testing continues to be controversial among experts and raises concerns among health care providers and regulatory agencies. The NIH-funded "Impact of Personal Genomics (PGen) Study" addresses these concerns by empirically measuring the perceptions and tracking the behaviors of individuals who have received DTC genetic testing from two separate companies. Research from this large-scale prospective study has already generated numerous new scientific reports. These findings shed light on who seeks testing and why, and how they respond to the results that they receive.

The latest results from the PGen Study were published on December 12 in the Journal of Clinical Oncology. This was an analysis of how customers respond to common cancer risk information (not Mendelian cancer risks like BRCA1/2), and was led by Stacy W. Gray, MD, City of Hope National Medical Center with senior author Robert C. Green, MD, MPH of Brigham and Women's Hospital, Broad Institute and Harvard Medical School. In this analysis, Gray and colleagues specifically looked at personal genomic testing for cancer risk, and found that 12-24% of individuals received "elevated" cancer risk estimates for prostate, breast and colon cancer. Despite learning they were at increased risk for these common cancers, most customers did not report changing their diet, exercise, supplement use, advanced care planning or cancer screening in comparison to the customers who learned they were at average or lower risk. The one exception is men who received elevated prostate cancer risk estimates as some of these men changed their vitamin and supplement use more than those at average or reduced risk. This counter-intuitive finding may have resulted because the increased cancer risks reported to the customers were very modest, and because the kind of individual who purchases DTC genetic testing may already have been very proactive about their health and using other available screening tests for cancer.

"These results suggest that people are not over-reacting to very modest cancer risks in DTC genetic testing" said Green, director of the Genomes2People Research Program, "and is consistent with some of our other findings showing that early adopters of DTC genetic testing

understand the limited predictive impact of DTC results and do not over-react either emotionally or in terms of generating additional and unnecessary medical expenses."

The advent in 2007 of DTC personal genomic testing, in which consumers could purchase genetic testing services directly from private companies, raised expectations for a new era of consumer genomics. This raised concerns about patients understanding the implications of their results and the response from doctors if asked to interpret these results. The PGen Study cohort is a group of more than 1,600 consumers who purchased personal genomic tests prior to the imposition of FDA restrictions in 2013 from 23andMe and Pathway Genomics (Pathway has since changed their business model and no longer provides DTC testing). In addition to sharing their actual test results with investigators, participants completed surveys before receiving their results and again two weeks and six months after receiving their results. The surveys also presented mock results to determine if consumers could accurately interpret them.

"There has been a tremendous amount of interest and opinion expressed about the potential benefits, harms and costs associated with personal genomic testing, and most of it has been speculative" said Green, "the PGen Study provided us with a goldmine of data on consumer expectations, how consumers interpret, recall and experience their results, how their results impact their state of mind, what actions they take after testing, and how all of these factors change over time."

"As far as many of the speculated risks and harms around direct-to-consumer genomic testing, we have not uncovered evidence that they are either common or severe," added Scott Roberts, PhD, of the University of Michigan's School of Public Health, and joint principal investigator of the PGen Study with Green. "Although we have found some areas where informed consent for testing and communication of results could be improved, our data suggest most consumers find their results as potentially useful in informing future health decisions and advance planning."

Here we provide a link to a complete list of scientific reports from the PGen Study. Some selected papers, along with their key findings from the PGen Study include:

- Ostergren et al. (2015) in *Public Health Genomics:* This study investigated **how well consumers understood** and could apply the information contained in their DTC genetic testing reports. Most consumers accurately interpreted sample test reports, but with some variation depending on consumer demographics (e.g., education level) and the type of results. For example, comprehension was lowest for carrier screening results, suggesting that companies could perhaps communicate these results more effectively.

- Meisel et al. (2015) in *Genome Medicine*: While the debates around DTC genetic testing have focused on its ability to predict future disease, this study demonstrated that consumers are particularly interested in genetic test results **that explain conditions they already have**, not just risks for diseases they may develop in future. This was found to be particularly true for conditions without an established cause.

- Baptista et al. (2016) in *Genetics in Medicine*: This study showed that **adopted persons who did not have access to their family histories valued DTC genetic testing even more** than non-adopted individuals.

- Kreiger et al. (2016) in *Nature Biotechnology:* Before-and-after surveys revealed that participants adjusted their assessment of their health risks by about twice as much when their results included **good news rather than bad news**. The study also found that consumers who were surprised by something in their results were the most likely to follow up by scheduling a doctor's appointment.

- Carere et al. (2016) in *Genetics in Medicine:* Comparison of consumers' real-world **prescription medication changes** with their personal genomic test results. The study found that while 5.6% of participants changed medications as a result of their genomic test results, only nine out of 961 participants - less than 1% - did so without first consulting a health care provider.

- Van der Wouden et al. (2016) in *Annals of Internal Medicine:* Six months after receiving their personal genomic test results, approximately 35% of participants had **shared their results** with their primary care physician or another health care provider. This was the first study to report consumer perspectives on those doctors' visits, with some interesting findings; for example, 22% of patients reported that their primary care physicians were dismissive of the genomic test results.

- Olfson et al. (2016) in *Nicotine and Tobacco Research*: DTC customers who were smokers show a high level of interest in genetic risks of smoking-related illnesses. The experience of receiving direct-to-consumer genomic health risks **did not lead to "false reassurance"** around their efforts to quit smoking.

This research was supported by the National Institutes of Health (NIH) National Human Genome Research Institute (Grant No. R01-HG005092); National Human Genome Research Institute (Grant No. U01HG006492); the American Cancer Society Grant No. 120529-MRSG-11-006-01-CPPB; a Canadian Institutes of Health Research Doctoral Foreign Study Award; and these NIH grants K07CA131103, U01-HG006500, U19-HD077671, U01-HG008685, and U41-HG006834.

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