At CGC, New PGen Study Data on Emotional and Practical Impacts of DTC Testing

October 09, 2013

By Molika Ashford

At last month's Consumer Genetics Conference, Robert Green of Brigham and Women's Hospital presented initial data from the PGen study, tracking subject's reported emotional reactions and follow-up behaviors to personal genetic testing up to six months after receiving results.

The data suggested that changes in mood and anxiety in the subset of people the study surveyed — early adopters of direct-to-consumer genetic risk testing — were transient: showing an initial shift at the time results were received, but then settling over six months back to normal levels. The data indicated that there was no difference in reported anxiety between subjects who had more elevated risks for various conditions versus those who had fewer. However, those who perceived themselves to be at greater risk based on their results had higher distress than others, regardless of their actual risk prediction.

In terms of the practical impacts of testing, Green reported at the conference that early analysis of the study data suggested that six months post-DTC testing, approximately 11 percent of study subjects had sought out some sort of follow up assessment or testing based on their genomic results with their doctors.

In an interview with *PGx Reporter* this week, Green acknowledged that it's hard to extrapolate from studies involving early adopters of DTC testing how the general population would respond to the risks and benefits associated with such testing.

"There are two layers of self-selection here," he said. "First, we have people who are, by definition, early adopters and who decided — or when offered the choice through an [online] portal like PatientsLikeMe, elected — to get personalized genetics testing as opposed to those who don't seek this out or don’t choose it when offered."

"Second, like with all studies, you have the volunteer bias of only studying those who choose to volunteer" for the trial, Green said.

There is always a possibility that those subjects who chose not to participate did so because they were more distressed by their results, or their experience with DTC genetic testing, than those who volunteered to participate in the study, Green explained. But, he said, "we were lucky [with PGen] in

that we didn't have too many who chose to turn us down." Green couldn't quantify how many people declined to participate in the study, but estimated that it was a "pretty small" percentage.

Regardless of how narrow a view studies of early adopters of DTC genetic testing might offer, Green said that the initial data coming out of the PGen analysis has yielded a number of interesting results in terms of who is being tested, and how they may be responding.

PGen, which kicked off in 2012 (*PGx Reporter 3/7/2012*), is a prospective analysis of the psychological and behavioral impact of personal genomics data, based on surveys of customers who received results from either 23andMe or Pathway Genomics.

In the study, researchers surveyed subjects at three time points: after they purchased a testing service through one or the other company but before they got results as a baseline measurement; then two-to-three weeks after they received their results; and finally six months after receiving results.

In his presentation at the Consumer Genetic Conference, Green said the demographic diversity of the study cohort turned out to be somewhat of a surprise. In an overall group of 1,848 respondents ,1,735 completed the baseline survey, about the same number viewed their test results, more than 1,100 participated in the post-results survey, and more than 1,000 responded to the final six-month survey.

"I expected Silicon Valley, super healthy, really high-income yuppies, but that wasn't really what we saw," Green said during his presentation at the conference.

Participants' ages ranged from 19-94 with a mean age of 48. Though the cohort was 90 percent white, 22 percent reported having below a college education; median income was $70,000 to $99,000 annually. The majority of subjects reported being in good to excellent health, while the other 15 percent considered themselves to be in fair to poor health.

Those who reported being in better health prior to testing tended to say their reasons for seeking testing were for fun and entertainment. Meanwhile, those who thought they were in worse health said they got tested to learn about specific disease risks and risks for their children out of a desire to improve health, for future planning purposes, and to gather information about family history, according to Green's data.

Participants were also surveyed about whether they had specific conditions prior to testing, including asthma, ulcerative colitis, obesity, bipolar disorder, and multiple sclerosis. The researchers asked how interested participants were in learning about their risk for diseases they already had versus those that they didn't, and found that people were most interested in risk information about the diseases they had.

"On one level, this is basic human nature," Green said at the conference. "On another level, it's profoundly irrational. This is a predictive paradigm for disease risk and certainly you don't need prediction for a disease you already have. But it may speak to a hidden agenda [in seeking this testing]. People are not only looking to these tests for predictive purposes, but also for explanatory purposes."

Almost all the people in the study reported that they would share their results with family during the baseline, pre-test survey, and about 90 percent said they had done so by the six-month mark. More than

50 percent of the participants said they would share their test results with friends, and even more reported doing so by six months. Sixty percent intended to share results with their primary care physician, and about half said they actually had done it six months later.

Only about three percent said they discussed results with a genetic specialist. "This may reflect paucity of genetic specialists or the fact that these people are essentially DIYers at heart," Green suggested in his presentation.

At the six-month mark, about 11 percent of the group said they had already made a doctor's appointment and another 10 percent intended to do so. Eleven percent said they had had tests, exams, or some other medical procedure based on their reported genetic information, including blood tests, eye exams, MRIs, CT scans, colonoscopies, and other genetic tests.

The potential impact of DTC genetic testing on healthcare utilization — whether it might rob the healthcare system of dollars — has been an area of great interest for industry observers.

"Depending on your perspective," he said at the conference, "that 11 percent may look like a high or low number. But if you multiply this by millions of people, if that is where this [testing] goes in the future, that could be a substantial pull on the healthcare dollar."

These findings have raised eyebrows among payors, according to Green. "I initially thought of that [11 percent] as a low number," he said. "But when I have talked with payors, they said they thought it was actually quite substantial."

According to Green, the study also revealed some interesting trends in subjects' perception of their genetic risk results. Those who perceived the results to be "interesting" were more likely to seek out follow-up care from a doctor, as were those who thought many or all of their results conferred high risk.

Previous studies of DTC test users, including research by the Scripps Translational Science Institute (*PGx Reporter 1/12/2011*) and the Multiplex Initiative by the National Human Genome Research Institute have shown that people who get tested through DTC genomics firms aren't necessarily more motivated to act on the results, but that the minority of people who do share their reports have appeared to be the most motivated to make lifestyle changes.

With regard to the emotional impact of DTC genetic testing, the participants in PGen who perceived their results as being most high-risk also reported higher anxiety than those who did not. But, this was not reflected in subjects' actual risk results. Those with a larger number of heightened risk results reported the same anxiety levels as those who had fewer elevated risk results.

"For those with more than 25 percent of conditions reported back as being at elevated risk, you would expect higher distress in this group. Yet, it looks virtually identical across the spectrum," Green said. "But it turns out that if you ask how many conditions they thought they had with increased risk, those who thought they had more had slightly, but statistically significantly, more distress."

Ultimately, the study shows that people who may be inherently more anxious tend to perceive test

results with more anxiety, according to Green. "What people walk in the door with in terms of risk anxiety and risk perception seems to have a very potent affect as a filter on how they see the information they are getting back and may even influence how they read their report," he added.

The study also showed that study participants' reported anxiety and depression over the course of the study showed a marked decrease and their positive mood showed an increase at the post-testing survey point from the pre-testing survey point, an then slowly rose or fell back to near pre-testing levels by the six-month mark.

"It's a very hard thing to disprove that anxiety or distress might kick in later, or long-after a person received DTC genomic results," Green told *PGx Reporter*. "But all of our data thus far really indicate the opposite: that any distress people feel is going to be in the short term and that people adjust very well as time goes by."

Green is also involved in a sequencing project involving newborns. He noted that the long-term impact of genetic test results is also of particular interest in this context "because of the notion that information given to a family at one stage of life could be more distressing at a later stage of life."

Green and his colleagues are still in the process of analyzing the full study dataset for PGen, doing both narrow analyses and wider treatments of the information they collected. In his presentation he mentioned that the group is specifically interested in drilling down and looking at specific risk categories and associated health interventions, such as BRCA mutation status, breast cancer testing, and treatment.

In a study published earlier this year by 23andMe an Stanford University, researchers found that among a cohort of women and men who chose to purchase testing from 23andMe and who learned that they were positive for certain BRCA mutations linked to a heightened risk of breast and ovarian cancer, none described feeling overly anxious (*PGx Reporter 2/20/2013*)