Eyeing 10K Enrollees by 2010, Coriell Institute Refines Personalized Med Portal for End Users

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Nearly halfway to meeting its goal of enrolling 10,000 people for its study looking at the impact of genomic-risk information on people's lives, the Coriell Institute updated researchers and industry representatives on the initiative at a conference on consumer genetics in Boston last week.

According to Erynn Gordon, senior genetic counselor at Coriell Institute, the personalized medicine project has so far enrolled 4,000 participants, and at this rate is hoping to meet its goal of 10,000 participants by 2010.

The project, called the Coriell Personalized Medicine Collaborative, or CPMC, was launched in December 2007 with the aim of studying the impact of genome-informed treatment on medical care. The study, which has partnered with Cooper University Hospital, Virtua Health, Fox Chase Cancer Center, and Helix Health, plans to ultimately enroll 100,000 participants.

In February the CPMC launched a web portal through which participants can answer questions about their family medical history and personal lifestyle. Factoring this information along with individuals' genotypes, CPMC will provide study participants with their risk for various actionable medical conditions, approved by its Informed Cohort Oversight Board.

The oversight board has so far approved the following conditions: Prostate cancer, type 1 and type 2 diabetes, age-related macular degeneration, melanoma, colon cancer, heart disease, iron overload/hemochromatosis, inflammatory bowel disease, and obesity. Enrollees to the study will receive their personalized risk information through the web portal by the second quarter of 2009, the CPMC states on its web site.

At the Consumer Genetics conference, Gordon presented a prototype of how genetic risk information would be displayed through the web portal and noted that after much debate, the CPMC decided to present this data in terms of a patient's risk of developing a disease relative to the general population, as opposed to a patient's absolute lifetime risk of developing the disease.

"We were reluctant to use absolute risk because that number is not fixed," Gordon noted, noting that algorithms for calculating absolute risk can be problematic and "as new gene risk variants are IDed [one's absolute] risk will change."

However, some at CPMC, including Gordon, felt that relative risk may be a difficult measurement for the public to comprehend. "As a genetic counselor, I was skeptical that people would understand relative risk," Gordon said, adding that most of the time, people just want to know if they have the genetic variant or not. However, since the project decided to use relative risk as a metric, Gordon noted she has been pleasantly surprised at how participants have digested the information.
Recently several commercial direct-to-consumer personal genomics firms developed a consensus document in which they agreed to use DNA chips with more than 99 percent accuracy covering between 600,000 and 1 million SNP markers; to characterize individual risk in terms of both absolute risk and relative risk; to use SNPs replicated in at least two well-powered studies; and to be transparent about their methods and criteria [see PGx Reporter 04-15-2009].

Since the recent launch of several DTC genetic screening services, industry observers have deliberated over the clinical utility of such information; raised ethical questions regarding marketing such information to consumers without much physician involvement; and debated the best way to present genetic risk information to the public.

According to Gordon, the CPMC is working with the US Food and Drug Administration to "figure out how to best present [genetic risk] information to the public in a responsible manner, so people don't start self medicating." Gordon did not elaborate on the nature of these discussions with the FDA.

Although the FDA has yet to express any intent to regulate the burgeoning DTC genetic testing industry, the FDA has said it may consider partnering with personal genomics firms in the future to use their large genetic risk association databases for post-marketing drug adverse events tracking [see PGx Reporter 11-19-2008].

"When you're DTC you have to be careful how you present this information," Gordon cautioned.

Separately, Courtney Sill, director of communications at Coriell Institute, told Pharmacogenomics Reporter this week that the CPMC is working with the FDA on a broad research collaboration looking at the pharmacogenomics of drugs. Sill could not provide any specifics on this project, but noted the collaboration with the agency would have "no direct regulatory implications."

Unlike commercial DTC consumer genomics firms, the CPMC is a non-profit research effort that doesn't charge participants for genotyping or risk analysis. Participants can choose to share their genetic risk data with their physicians through the web portal. The CPMC currently has $5 million in funding, drawn from private philanthropy, foundation grants, and institutional support.

Gordon noted that the CPMC is exploring different research collaborations with pharmaceutical companies but has not yet inked any such partnerships. "We want to make sure this data is available to academics and industry," Gordon noted. "We have talked about serving as a middleman linking [CPMC] study participants with pharma-run clinical trials," but there has not been anything solidified in this regard.