Growing Use of Genomic Data Reveals Need to Improve Consent and Privacy Standards

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In March, the publication of the complete genome sequence of cancer cells from a Maryland woman who died in 1951 ignited an ethical firestorm. These cells, called HeLa because they were derived from the cervical tumor of Henrietta Lacks, have been widely cultured in laboratories and used in research.

As documented in a 2010 book, the cells had been taken for research use without consent, and for years the woman’s family had been largely unaware of their use (Skloot R. The Immortal Life of Henrietta Lacks. New York, NY: Crown; 2010). The publication of the sequence (Landry JJ et al. G3 [Bethesda]. doi:10.1534/g3.113.005777 [published online March 11, 2013]), again without the consent of Lacks’ family, renewed debate about the privacy and security of genomic information in research, as Rebecca Skloot noted in the New York Times (http://tinyurl.com/c8wgp3f). By late March, the authors removed the sequence from an online database at the request of the family.

As the Presidential Commission for the Study of Bioethical Issues noted in a report last fall (http://www.bioethics.gov/cms/sites/default/files/PrivacyProgress508.pdf), the proliferation of genomic sequence data in research and its great promise in the clinical realm demand a coordinated effort to protect the privacy of individuals and their families when genomic data are used in research. Doing so is essential to ensure that enough individuals are willing to participate in such research to allow continued scientific progress, said Lisa M. Lee, PhD, MS, executive director of the commission.

“We need people to trust that these data will be protected,” Lee said.

Lee explained that advances in sequencing technology and rapidly declining costs of sequencing are likely to accelerate the use of genome sequences in research and in the clinic, but that policies on such data haven’t yet caught up with the science. For instance, she noted that a genome sequence acquired by a physician in the course of treating a patient would be protected under the Health Insurance Portability and Accountability Act, but if the same sequence were collected by researchers, it would fall under the so-called Common Rule. Additionally, the commission’s report cites a patchwork of varying laws from state to state. For example, in some states it is legal to surreptitiously collect and sequence DNA from an unwitting individual, such as by extracting DNA from saliva on a discarded coffee cup.

The commission recommended the enactment of a consistent floor of protections across the states. Lee said the commission did not stipulate what these protections should be or how they should be created, but that a separate process should establish them. The report also recommends that research funders, researchers, and those housing genomic data establish standards for appropriate use and security of the data, including options for patients who would want to share their data widely.

The report also calls for the creation of a robust consent process for research, with guidelines from the Office for Human Research Protections. According to the report, consent should include the following:

- A description of sequencing and genome analysis
- How the data will be used now and in the future
- The degree to which participants can control future use of the data
- The benefits and potential risks of participation, including the potential for unknown future risks
- What data and information might be returned to the participant, such as whether they will be informed of incidental findings

One unique feature of genomic information that patients must understand is the potential for the data to reveal information about family members, Lee said.
"We are learning about our parents', siblings', and future children's genomes," she said. "We need to be clear with people about what this may mean."

Anne Townsend, PhD, research associate in medical genetics at the University of British Columbia in Vancouver, Canada, said she was pleased that the guidelines were driven by respect for individuals. She emphasized that proper informed consent is crucial, including consent regarding whether incidental findings will be returned.

One of several vignettes included in the report highlights a young man who was diagnosed with retinitis pigmentosa at age 13 years and told he would lose his sight at some point later in life. The information drove the man's career choices and created emotional distress in the years leading up to blindness. The man later chose to participate in a genome sequencing study, but only on the condition he not be notified of incidental findings.

"One of the downsides of knowing about a future health outcome is that it could cast a shadow over our lives," Townsend said.

Townsend said such personal stories are useful and that other research has suggested that, contrary to what some might anticipate, many individuals with genetic conditions such as Huntington disease do not want to know in advance. She said that because people assess risk in different ways, it's important not to make assumptions about how people would handle information or about what they would choose.

Townsend said that more creative and interactive ways of obtaining consent using new technologies may help patients give more informed and meaningful consent about participating in genetic research. She noted that interactive patient decision-making tools are already being used in other areas of medicine. More study of genomic research participants' thoughts and experiences is also needed to provide a rigorous evidence base for developing consent processes.

One weakness of the report, she said, was that it didn't pay enough attention to potential blurring of lines between patients and participants. For example, she noted that patients may need greater protections because they may be more vulnerable. For example, a patient may see participating in a research study as a means to access care.

Lee said that creating the appropriate protections for individuals involved in genomic research is crucial if society hopes to reap the potential benefits of genomics. "Without appropriate protections, we can't progress scientifically," she said.

### Physicians, Patients Not Following Advice From USPSTF on Mammography Screening

Mike Mitka, MSJ

In 2009, the US Preventive Services Task Force (USPSTF) recommended against routine screening mammography for women aged 40 to 49 years and advised biennial rather than annual screening for women aged 50 to 74 years. But it appears that women and physicians have ignored these recommendations.

A new study by researchers from Harvard Medical School and Brigham and Women's Hospital in Boston found that in 2005, 2008, and 2011, the percentage of women aged 40 to 49 years reporting that they had undergone mammography screening in the previous year was the same, about 47%. As for women aged 50 to 74 years, the percentage reporting mammography screening in the previous 12 months for each year analyzed also remained essentially the same, in the upper 50% range.

The researchers based their findings on data from 27,829 women aged 40 years and older who participated in the 2005, 2008, and 2011 National Health Interview Surveys (Pace LE et al. Cancer. doi:10.1002/cncr.28105 [published online April 19, 2013]).

The USPSTF's rationale for its 2009 recommendations was that for women aged 40 to 49 years who are at relatively low risk for breast cancer, false-positive results are more common and result in psychological harms, unnecessary imaging tests, and biopsies in women without cancer. As for women aged 50 to 74 years, too-frequent screening could lead to overdiagnosis, resulting in treatment of cancers that would not become clinically apparent during a woman's lifetime (http://tinyurl.com/3cxvx9y). Other research finds that although there has been a substantial increase in the number of cases of early-stage breast cancer detected over the past 3 decades by screening mammography, there has been only a marginally reduced rate at which women present with advanced cancer (Bleyer A and Welch HG. N Engl J Med. 2012;367[21]:1998-2005).

When released, the task force's recommendations sparked a great deal of criticism by members of certain advocacy, primary care, and oncology communities, criticism that was widely reported in media stories about the issue. The Cancer researchers suggest that this criticism and media coverage may have discouraged adoption of the task force recommendations.