



Technology: The ethical and legal challenges of large-scale human genome sequencing

The progression of genetic technology has spawned security, privacy and ownership concerns

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The Presidential Commission for the Study of Bioethical Issues recently issued a request for public comment on a variety of issues raised by large-scale human genome sequencing, and just last week The New York Times [ran an article](#) on Washington University's effort to save the life of Dr. Lukas Wartman, who was diagnosed with leukemia, by fully sequencing the genes of his cancer cells and his healthy cells for comparison. After finding that Dr. Wartman had a normal gene that worked in overdrive, spurring growth of his cancer, researchers were able to identify and use a new drug—which had been tested and approved only for advanced kidney cancer—to shut down his malfunctioning gene. Researchers say that Dr. Wartman's case is an illustration of the importance of genes in driving a cancer, as opposed to the organ or tissues where the cancer originates.

Whole genome sequencing, along with greatly improved DNA sequencing techniques, has the potential to improve our understanding of the pathogenesis of disease and enhance drug design. Many predict that within a decade or so whole genome sequencing will be widely available at a price of \$1,000 or less and perhaps paid for by insurance. Industry and venture capitalists are increasingly interested in initiatives to identify specific disease-causing genes and to test drugs that target those genes.

A number of ethical and legal issues present challenges to this approach. One issue relates to ownership of and control over the tissue sample that contains the DNA and derivative information. The court in *Moore v. Regents of the University of California*, a leading case on the topic, rejected the notion that Moore had a property right to his spleen cells that were removed during a splenectomy and then commercially exploited. Rather, the court held that Moore had an informed consent right to be told by his physician about the planned removal and use of his cells and then to decide whether to provide them.

A few states, however, have enacted or are considering laws that would explicitly establish individuals' ownership rights to their genetic samples. For example, in Alaska, a DNA sample and the results of DNA analysis performed on the sample are the exclusive

property of the person sampled or analyzed. The difference in approach to this legal ownership question can be explained, in part, by the fact that today, unlike 20 years ago when *Moore* was decided, DNA sequence variants can readily serve as a source of personal identification, and that it is even possible to re-identify individuals in pooled mixtures of DNA. If tissue samples that contain DNA are not de-identifiable, legitimate concerns arise about previous practices that accommodated research use of discarded tissue without the consent or even knowledge of the individuals from whom the samples were extracted.

Laws that explicitly establish individuals' ownership rights in their tissue in order to guard against these concerns, however, have important implications for research involving large-scale human genome sequencing. Some fear that such laws could have a chilling effect on the development of biorepositories for research purposes, due to fear of private claims for violation of individuals' property rights in their DNA samples. Some worry that procedures for transfer of an individual's property right in his samples to a research entity will run afoul of legal and ethical concerns about selling human tissue. And some hypothesize that individuals will have a greater ability to place limitations on the research use of their samples.

Rapid technological advances complicate not only tissue sample ownership but also the use of genetic information obtained via genome sequencing performed with consent of the tissue donor. In order to protect autonomy and privacy interests, it is critical to obtain fully informed consent from the tissue donor for whole or partial genome sequencing, subsequent testing and use and disclosure of test results. The informed consent discussion must address:

- Risks, which depend on what information the genome sequencing reveals
- The personal and social significance of that information
- Security protections for the DNA and genotypic information
- The types of data misuse that could occur

For instance, one risk of whole genome sequencing is that the individual might learn more information about himself and his family than he prefers to know or have known by others, especially if such information could lead to embarrassment, stigmatization or discrimination. In addition, adequate legal protection of the privacy of genetic information is increasingly important as genetic technology progresses. The 2008 Genetic Information Nondiscrimination Act, which prohibits use of genetic information in health insurance and employment matters, provides an important starting point. Unless legal protections keep pace with evolving technologies, however, public confidence in the promise of genomics to improve health will be at risk.