



## FROM BIRTH TO DEATH AND BENCH TO CLINIC

# THE HASTINGS CENTER BIOETHICS BRIEFING BOOK

for Journalists, Policymakers, and Campaigns

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### CHAPTER 3

## Biobanks: DNA and Research

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# biobanks: dna and research

by Karen J. Maschke

## Framing the Issue

With recent advances in molecular biology, human biospecimens have become enormously valuable for medical researchers. Biospecimens such as blood, surgical tissue, saliva, and urine contain genetic material that researchers analyze to identify gene variations associated with human diseases. By identifying the role that genes play in disease formation, researchers may be able to develop new diagnostic tests and targeted treatments for specific diseases and to investigate how genes interact with environmental factors. This research may also open the way to “personalized medicine”—treatments that are customized to a person’s genetic profile (see chapter 29, “Personalized Medicine and Genomics”).

By 1999, over 300 million biospecimens were stored in the United States in a wide variety of public and private repositories. Most of these biospecimens were collected during routine clinical and surgical procedures.<sup>1</sup> Over the last decade, several new public and private initiatives in the United States and elsewhere have been collecting and storing biospecimens for research purposes. For instance, in 2003, seven disease organizations in this country created the Genetic Alliance BioBank, an initiative that collects, stores, and distributes biospecimens and clinical data to researchers studying certain diseases. More recently, Children’s Hospital of Philadelphia launched a project to collect blood samples from over 100,000 children for the purpose of conducting genetic research on the most prevalent diseases of childhood. The best known initiative outside the United States is the UK Biobank, a public-private partnership currently recruiting 500,000 people in the United Kingdom aged 40 to 69 years to donate blood and urine samples for long-term storage. Researchers will use these biospecimens, data derived from them, and data provided by individuals at the time of enrollment to study the correlation between genetics, environment, and lifestyle in disease formation.

Although the procedures for collecting biospecimens and extracting genetic information from them are relatively straightforward, the ethical, legal, and social challenges associated with biospecimen research are not so clear-cut. These challenges include matters involving informed consent, privacy and confidentiality, disclosure of research results, intellectual property, and biobank governance.

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## HIGHLIGHTS

- More than 300 million biospecimens are stored in the United States in public and private repositories known as biobanks.
- These biospecimens—human blood, surgical tissue, etc.—contain genetic material that can be analyzed to identify gene variations associated with human diseases.
- These gene variations may lead to new diagnostic tests and targeted treatments for specific diseases, as well as open the way to personalized medicine.
- Collecting and storing biospecimens raises ethical challenges concerning how to obtain informed consent, protect privacy, and disclose research results.
- Other issues concern the ownership of biospecimens as intellectual property and government oversight of biobanks to ensure ethical use of biospecimens.

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## Consent, Risk, and Privacy

There is international consensus that for most research with humans to be ethical, at least two key requirements must be met: an ethics review board—known as an institutional review board in the United States—must review and approve a study before researchers recruit participants, and participants must voluntarily consent to be in the study. The purpose of ethics review is to ensure that persons independent of the research determine that the study's potential benefits to participants outweigh the potential risks of research participation. The informed consent rule is to ensure that individuals who enroll in a study understand its purpose and voluntarily agree to expose themselves to potential research risks.

Applying the informed consent rule to research with biospecimens is problematic for several reasons. Because many stored biospecimens were collected for purposes other than research—sometimes during routine clinical and surgical procedures—individuals did not give consent for those biospecimens to be used in research. In other instances, individuals may have given consent for specific types of research with their biospecimens, but later on researchers want to use them for other types of studies. Additional complexities surrounding the consent issue have to do with the use and disclosure of genetic and other identifiable medical data.

There are also ethical challenges in obtaining consent when biospecimens are collected. Individuals who provide biospecimens to the UK Biobank give blanket consent for research with their biological materials. This means that no restrictions are placed on the types of research that can be conducted with their biospecimens. Some commentators contend that blanket consent doesn't meet the definition of informed consent because individuals do not have full information about how their biospecimens will be used.

One alternative is tailored consent, which gives individuals a choice about the specific types of research for which their biospecimens and related information can be used. For instance, an individual providing a biospecimen for research might authorize cancer research with the biospecimen but not diabetes research. The Genetic Alliance BioBank, which uses tailored consent, has a process that lets researchers recontact individuals to obtain consent for new research with their stored biospecimens as the need arises. And in its recently pub-

## WHO OWNS BIOSPECIMENS?

*Institutions with biobanks in this country usually claim ownership rights over biospecimens. Legal challenges to this claim have been unsuccessful. Here is a sampling of significant cases.*

- **Moore v. Regents of the University of California** – In 1990, a California court ruled that individuals do not have an ownership interest in their cells after the cells have been removed from their bodies.
- **Greenberg v. Miami Children's Hospital Research Institute, Inc.** – In 2003, a federal district court in Florida ruled that individuals do not own their tissue samples.
- **Washington University v. Catalona** – In 2008, the U. S. Supreme Court declined to review a biospecimen ownership case. The question was whether individual donors who provide biospecimens for research “retain an ownership interest allowing [them] to direct or authorize the transfer of such materials to a third party.” The court of appeals said, “the answer is no.”

lished “Best Practices for Biospecimen Resources,” the National Cancer Institute permits, but does not require, the researchers it funds to use a tiered consent process if appropriate for the study's design or the biobank's mission. With the tiered process, human subjects could specify the types of research for which their biospecimens could be used.

Many commentators contend that the risk of harm from research with biospecimens is low and primarily related to the disclosure of a person's identifiable genetic and other medical information. Thus, some claim that blanket consent is ethically acceptable when biospecimens are collected, and that consent for secondary uses is not required if there are adequate safeguards to protect the privacy and confidentiality of identifiable medical information. Proposed safeguards include requiring ethics review boards to approve new studies with stored biospecimens and associated data, deidentifying biospecimens and associated data with no means to relink them to identifiable persons, establishing rules for relinking deidentified biospecimens and associated data to identifiable persons, and establishing security measures to minimize unauthorized access to biospecimens and associated data.

Because the United States has no comprehensive regulatory framework that addresses these issues, there is confusion about when the rules governing research with humans apply to research with biospecimens and their data. Some rules con-

flict with each other, and there are differences in how the rules define “research,” “human subjects,” and “identifiable” personal information. How these terms are defined determines whether IRBs must approve biospecimen research, and whether individuals must give consent for use of their stored biospecimens or their identifiable genetic and other medical information. Not all state genetic privacy laws apply to research with genetic data, and the federal Health Insurance Portability and Accountability Act (HIPAA) privacy rule may or may not apply to identifiable genetic and medical information associated with biospecimens.

## Disclosing Research Results

Not all research results are published, and even those that are usually appear long after the study began and in specialized science journals. Thus, people who provide biospecimens for research—along with the general public—might never learn the outcome of that research. There are reasons for not disclosing some findings. Preliminary or inconclusive results might not have clinical value and could provide misleading information about the causes of disease, especially when the research involves analysis of genetic materials. Genetic research results have privacy implications for family members who did not provide biospecimens for research. Moreover, most genetic tests on human biospecimens are conducted in laboratories that don’t meet quality standards under the Clinical Laboratory Improvement Amendments of 1988 (CLIA). Only CLIA-certified laboratories are permitted to disclose test results for clinical decision-making.

Despite these concerns support is growing for the principle that researchers and study sponsors have an obligation to disclose both positive and negative research results, provided that certain conditions are met. Yet how and when to do this remains unclear. A working group of the National Heart Lung and Blood Institute recently recommended that researchers disclose results of genetic studies when the associated risk for the disease is significant, when the disease has important health or significant reproductive implications, and when proven therapeutic or preventive interventions are available. The exceptions to this recommendation are genetic-related diseases such as Huntington disease that, although untreatable, have reproductive implications.

## RESOURCES

### Web sites

- [www.primr.org](http://www.primr.org) – Public Responsibility in Medicine and Research. Resource Center includes abstract and article archives, best practices and standard operating procedures, and other publications exploring the ethical issues of research with biospecimens.
- [bioethics.georgetown.edu/nbac/](http://bioethics.georgetown.edu/nbac/) – the National Bioethics Advisory Commission. Reports page includes several relevant publications under “Research Involving Human Biological Materials: Ethical Issues and Policy Guidance.”

### Recent news

- Anita Huslin, “Firm Carves New Model in Biotech Research,” *Washington Post*, December 17, 2007.
- Colin Nickerson, “Gene Advances Bring Ethical Quandaries,” *Boston Globe*, May 11, 2007.

### Further reading

- National Cancer Institute, “Best Practices for Biospecimen Resources,” June 2007. Report available at <http://biospecimens.cancer.gov/practices/>.
- Public Responsibility in Medicine and Research, “Report of the Human Tissue/Specimen Banking Working Group, Part I: Assessment and Recommendations,” March 2007. Report available at [www.primr.org](http://www.primr.org).
- Robert F. Weir and Robert S. Olick, *The Stored Tissue Issue: Biomedical Research, Ethics, and Law in the Era of Genomic Medicine*, Oxford University Press, 2004.

## Ownership and Intellectual Property

The UK Biobank’s ethics and governance document explains that the biobank owns the stored biospecimens, as well as the data stored in the biobank that came from the biospecimens, from the individuals’ medical records, and from the individuals themselves when they provided information at the time of enrollment. The governance document also says that individuals have no property rights to their stored biospecimens.<sup>2</sup>

Institutions in the United States also typically assert ownership rights over biospecimens stored in their repositories. But some commentators—including researchers and individuals who provided biospecimens for research—have challenged this ownership claim. To date, challenges have been unsuccessful.

Because research with biospecimens and associated data may lead to inventions that have commercial value, the impulse for research sponsors, researchers, and institutions with repositories is to

use intellectual property rights to control access to those resources. Policies regarding intellectual property rights vary. The NCI's "Best Practices for Biospecimen Resources" says that researchers and institutions should share research data and tools generated through use of biospecimens in a timely manner, and that biorepositories have no inherent rights to future intellectual property, such as reach-through rights to inventions made by using repository samples.

## Policy to Govern Biobanks

Who gets access to biospecimens and associated data? What, if any, conditions should be placed on the use of these resources? Who makes these decisions? These are important policy issues. Although the public supports research with biospecimens, there is concern that they may be used for research that some find objectionable, like cloning, and that genetic and other medical information may be used in ways that can harm individuals and their families, such as stigmatizing them because of a genetic condition. Widespread fear that employers might use genetic information to deny people

employment might be allayed now that President Bush has signed the Genetic Information Nondiscrimination Act (GINA), which prohibits employers and health insurers from discriminating against individuals on the basis of their genetic information.

One governance model is the UK Biobank's Ethics Governance Council, an independent committee that provides advice to the UK Biobank and monitors its activities to ensure that it conforms to policies laid out in the Ethics Governance Framework. Whether this type of governance mechanism or different ethics frameworks used by other biobanks will be effective remains to be seen. 

## References

1. E. Eisman and S.B. Haga, *Handbook of Human Tissue Sources: A National Resource of Human Tissue Samples* (Santa Monica, CA: RAND, 1999).
2. UK Biobank Ethics and Governance Framework. Version 3.0, October 2007, at <http://www.ukbiobank.ac.uk/docs/EGF20082.pdf>.