Presidential Commission *for the* Study of Bioethical Issues

Community Engagement in *Privacy and Progress in Whole Genome Sequencing*

Contents

I. Introduction

In *Privacy and Progress in Whole Genome Sequencing* (*Privacy and Progress*), the Presidential Commission for the Study of Bioethical Issues (Bioethics Commission) highlights an important tension regarding whole genome sequencing: how to reconcile the potential for important medical benefits for society at large with the privacy interests of individuals who choose to share their whole genome sequence data.[1] The report assesses the challenges that face the medical and research communities as whole genome sequencing technology becomes less expensive and more prevalent.

II. Learning Objectives

Students should be able to:

1. Describe the importance of engaging communities in the sharing of whole genome sequence data.

2. Explain how community engagement helps advance the ethical principles articulated in *Privacy and Progress*.

[1] Presidential Commission for the Study of Bioethical Issues (PCSBI). (2012, October). *Privacy and Progress in Whole Genome Sequencing*. Washington, DC: PCSBI.

3. Identify ways in which community engagement might facilitate the recommendations made in *Privacy and Progress*.

III. Background

Privacy and Progress was framed around five ethical principles that should be considered in evaluating emerging technologies such as whole genome sequencing: 1) public beneficence, 2) responsible stewardship, 3) intellectual freedom and responsibility, 4) democratic deliberation, and 5) justice and fairness. These principles, in addition to the enduring principle of respect for persons, which sets the foundation of privacy, are outlined in the introduction of the report, on pages 28-30. The highlighted principles support the use of community engagement, especially the ethical concept of democratic deliberation, described in the introduction:

> Democratic deliberation is an approach to collaborative decision making that embraces respectful debate of opposing views and active participation by citizens. Democratic deliberation warrants engaging the public and fostering dialogue among the scientific community, policy makers, and persons concerned with the issues raised by scientific progress.[2]

The report's first recommendation, Recommendation 1.1, emphasizes the principle of democratic deliberation, and supports the idea of community engagement.

> **Recommendation 1.1**
>
> **Funders of whole genome sequencing research; managers of research, clinical, and commercial databases; and policy makers should maintain or establish clear policies defining acceptable access to and permissible uses of whole genome sequence data. These policies should promote opportunities for models of data sharing by individuals who want to share their whole genome sequence data with clinicians, researchers, or others.[3]**

Recommendation 1.1 articulates a need for researchers and policy makers to establish clear policies for acceptable access to and use of genomic information. In explaining the importance of that first recommendation, the Bioethics Commission stated:

> Developments in the science of whole genome sequencing, which are progressing quickly, will require ongoing ethical consideration and

[2] Ibid, p. 30.
[3] Ibid, p. 76.

> democratic deliberation. Individuals and groups have differing sensibilities toward the privacy and publicity of whole genome sequence data, which might be relevant to distinguishing between acceptable and unacceptable uses of data. Perceived misuses of whole genome sequence data vary between cultures and individuals. For example, some individuals might be open to having a secondary researcher use his or her whole genome sequence data for an ancestry study. Members of the Havasupai tribe, on the other hand, strongly disapproved of their samples being used in ancestry studies, because these studies contradicted their traditional origin beliefs. Some parents do not object to using Guthrie card newborn blood screening spots in future research without consent. Notable lawsuits in Minnesota and Texas, however, have indicated that some parents feel otherwise.[4]

In *Privacy and Progress*, the Bioethics Commission emphasized the potentially sensitive nature of genomic data and the information that flows from it. Genomic data can be collected through biological samples and stored with related medical information in biobanks (or biorepositories) for use in current or future research in the clinical, research, or direct-to-consumer settings. As a result, the community of participants involved in biobanking research might be difficult to define, identify, engage, and re-contact. Given individuals' differing backgrounds, cultural sensitivities, and wide range of opinions, community engagement is salient as a method for eliciting and acknowledging such plurality of values in genomic research.

In addition, the community engagement process demonstrates several other ethical principles articulated by the Bioethics Commission, including the overarching principles of respect for persons, responsible stewardship, and justice and fairness. Respect for persons requires that researchers safeguard the autonomy of research participants. Community engagement can lead to tailored informed consent procedures specific to a designated community that incorporate community ideals, acknowledge community values and concerns, and ensure full and understandable explanations of the risks and potential benefits of the research to allow for fully autonomous decision making on the part of participants. Listening to community ideas, being sensitive to community concerns, and incorporating community feedback in decisions about study design, recruitment, and dissemination of results also demonstrates an application of the principle of respect for persons at the community level.

Responsible stewardship requires that we take into account the needs of those who cannot represent themselves, such as children, future generations, the mentally incapacitated, or

[4] Ibid, p. 75.

groups that might be affected by, but remain unaware of, the risks. Community engagement can aid in this pursuit by identifying stakeholders, ensuring that representatives have the opportunity to be heard, and eliciting how whole genome sequencing comports with the stakeholders' specific needs. Due to the heritable nature of genomic information, it is especially important for researchers and clinicians to employ the principle of responsible stewardship to consider the voices of future generations; garnering the opinions of communities of present generations can help achieve this goal.

Finally, the principle of justice and fairness calls upon the scientific community to assure that the burdens and benefits of whole genome sequencing do not fall disproportionately on any particular group. Community members provide knowledge of community relationships and practices which can inform predictions of the consequences of genomic research. Such knowledge helps researchers to anticipate previously unforeseen harms and benefits and helps to structure research design, conduct, and dissemination of results accordingly. Listening to and incorporating input from the community can help ensure justice and fairness in the use of whole genome sequencing technologies.

Privacy and Progress highlights the particular privacy concerns that individuals face when deciding whether to allow access to or use of their whole genome sequence data. However, decisions that individuals make can have implications for entire communities or populations, especially in this particular realm of science. For example, genetic screening for Tay-Sachs disease in Ashkenazi Jews must ensure respect for the privacy of those tested and the Ashkenazi Jewish community should be protected against potential stigmatization and discrimination.[5] As such, engaging communities whose members might consent for researchers, doctors, or commercial entities to use their whole genome sequence data can ensure that potential implications for populations, groups, and individuals are taken into account before data are used.

Careful consideration of the input from the community of whole genome sequencing participants, implicated populations, and other stakeholders, and thoughtfully informing them of potential research parameters, benefits, and risks are all important pieces of achieving appropriate community engagement in accordance with the ethical principles discussed in *Privacy and Progress*.

[5] Khoury, M.J., et al. (2003). Population screening in the age of genomic medicine. *New England Journal of Medicine*, 348(1), 50-58.

IV. Reading

For the purposes of discussion, have students download and read the following Bioethics Commission materials (reports can be found on the Bioethics Commission website at www.bioethics.gov under "Projects"):

> *Privacy and Progress in Whole Genome Sequencing*, pp. 13-32 ("Introduction").
>
> *Privacy and Progress*, pp. 72-77 ("Recommendation 1.1").
>
> *Privacy and Progress*, pp. 91-93 ("Recommendation 3.1").
>
> *Privacy and Progress*, p. 130 ("Note 68").

V. Discussion Questions

The following questions are based on the information provided above and through the indicated reading and are intended to reinforce important aspects of community engagement as it relates to whole genome sequencing and the Bioethics Commission's *Privacy and Progress* report. Important points are noted with each question to help the instructor guide a group discussion. The "Additional Reading" section will be helpful in answering these questions.

1. **Engaging the community in decisions to share whole genome sequence data can be more difficult than community engagement in other types of medical or research decision making. What is distinct about whole genome sequencing that makes this the case?**

   Starting points for discussion:

   a. *Complexity*: Genome science is complex and the implications of deciding to share one's whole genome sequence data can be equally difficult to understand, especially when one considers the implications of as yet undetermined future research on stored genomic samples or data.

   b. *De-identification/anonymization*: Whole genome sequencing data are often de-identified and anonymized, coding or removing the link between an individual and his or her data so that a researcher cannot know to whom the data belong. This also weakens the ties between data and the community, but in studies of specific biologically linked ethnic or cultural groups, there is still a danger of stigmatization despite the de-identification

or anonymization of individual data.[6] This is because information about one individual could also reveal information about a group.

c. *Benefit and risk*: Genomic studies generally do not result in direct benefit to participants; rather, they lead to potential future public benefit. However, the risk incurred by sharing one's whole genome sequence data is attributed only to the individuals or communities that share their data.

d. *Defining community*: Collection of DNA or other biological samples for biobanking can occur in the clinical, research, or direct-to-consumer settings. As such, the community of participants involved in biobanking research might be difficult to define, identify, engage, or re-contact.

2. How can community engagement help foster more ethical and culturally sensitive access to and use of genetic and whole genome sequence data?

Starting points for discussion:

a. Community engagement is important in whole genome sequencing because genetic and genomic data are intimately tied to individuals' identity and sense of self. As such, input from potentially affected community members is valuable to ensure that data and results are handled and presented in a sensitive and respectful way. The sharing of whole genome sequence data for research can ultimately serve to sharpen or to blur cultural, racial, and national differences, and therefore requires handling that is sensitive, empathetic, and supported by communities.

b. *Havasupai case:* Read about the case of the Havasupai tribe on page 130, note 68 of *Privacy and Progress*, which demonstrates and highlights the role of cultural sensitivities in genetic and genomic research. This case illustrates why community input is so important. Note: a more thorough analysis of this case is described in Scenario A of the "Problem-Based Learning" section of this module.

3. What special issues should researchers consider when informing communities about the risks and benefits of whole genome sequencing research?

Starting points for discussion:

[6] An article published after *Privacy and Progress* in the journal *Science* suggested that genomic data can never be fully de-identified. See Gymrek, M., et al. (2013). Identifying personal genomes by surname inference. *Science*, 339(6117), 321-324; See also Gutmann, A. (2013). Data re-identification: Prioritize privacy. *Science*, 339(6123), 1032.

a. *Need for data*: By analyzing large numbers of genomes, scientists can identify genetic variants that might be linked to diseases, which can then be studied for potential treatments. In order to secure such benefits and advance medical understanding for the public good in this way, researchers require access to large numbers of whole genome sequences coupled with associated medical information.

b. *Privacy*: Researchers should acknowledge the privacy concerns raised by the generation, use, and storage of whole genome sequence data.

c. *Transparency*: Researchers can engage the community that might choose to participate in whole genome sequencing research through frank discussions of the goals of prospective research, how the collective datasets will be stored for use in the future, whether results will be shared or publicly available, and potential implications of future use of the data. In addition, although individuals can make their own choices about whether to participate in the research, those decisions might have implications for communities or populations as a whole if data are shared. As such, community engagement can help address that concern and inform individuals of the potential implications of their decisions.

4. **In *Privacy and Progress*, the Bioethics Commission explains that whole genome sequence data obtained in the clinic can later be anonymized and used in studies by researchers within the same institution and by future researchers once such data are shared. Under these circumstances, how might community be defined? How can those who handle whole genome sequence data seek and take into account guidance provided by the community?**

   Starting points for discussion:

   a. *Informed consent*: See Recommendation 3.1, regarding consent for whole genome sequencing research, and a discussion of how the informed consent process can address some of these points and concerns with individuals (*Privacy and Progress*, pp. 91-93).

   b. Consider how engaging the patient/participant community, and larger communities that might be affected by genomic research, during the planning stages of research can improve informed consent processes and lead to increased understanding of whole genome sequencing research, demonstrating the ethical principle of respect for persons.

c. Consider mechanisms beyond the informed consent process that might engage the larger community (e.g., a dedicated information resource phone line or public meetings).

5. In *Privacy and Progress*, the Bioethics Commission explains the continuum between identifiable, de-identified, and anonymized whole genome sequence data (pp. 62-65). How should community engagement be employed when researchers use de-identified and anonymized data? If the community of individuals who donated the genomic material cannot be contacted, what strategies might researchers employ to engage the community?

Starting points for discussion:

a. Identifiable data, those that can be linked to an individual, are covered by federal regulations designed to protect research participants. These regulations can be found in the U.S. Department of Health and Human Services Code of Federal Regulations at 45 C.F.R. Part 46, (Subpart A of which is often referred to as the Common Rule).

b. Use of de-identified data for research, however, is not considered research with human subjects and therefore is not subject to the protections outlined in the Common Rule (*Privacy and Progress*, pp. 64-65). Community engagement with groups of research participants can lead to greater clarity in informed consent processes when specimens are initially collected.

c. For data that are generated through the clinic, community engagement with patient groups can lead to improved clarity in informed consent processes, regardless of whether data are de-identified at a later time.

d. Engaging the community while planning the research design enables researchers to seek the community's guidance regarding whether data should be de-identified, or stored in a data bank, and, if so, how future research using the data should be handled. This would be addressed in the planning stages and would be specified in the informed consent process. Since de-identified data are less strictly regulated, does this suggest that community engagement to attain ethical research practices might be more important for de-identified whole genome sequencing studies? Consider this and discuss.

VI. Problem-Based Learning

Scenario A. *In the 1990s, Arizona State University (ASU) researchers collected DNA samples from members of the Havasupai Native American tribe to explore potential genetic links to the high rate of diabetes in the Havasupai population. Members of the tribe claimed researchers shared the samples with other researchers without obtaining consent for further research including studies involving mental illness and theories regarding the tribe's geographical origins that contradict their traditional beliefs, studies that the tribe members found personally and culturally offensive. Although this example describes research using discrete genetic tests rather than whole genome sequencing—that is, a specific targeted test rather than a survey of the entire genome—many of the same principles apply.*

> Transcripts and archived webcast video of Carletta Tilousi's presentation to the Bioethics Commission on Tuesday, August 30th, 2011 are available on the Bioethics Commission's website under Meeting 6, Session 5 (beginning at 03:17 on the webcast video).

Have students watch Ms. Tilousi's presentation and discuss the following:

1. **How might engagement with the Havasupai community have helped obviate concerns about research conducted at ASU and other institutions?**

   Starting points for discussion:

   a. The Havasupai tribe members would have had the opportunity to articulate their values, desires in participating in research, and ideal outcomes. Additionally, they might have been able to express preferences regarding sharing of samples and data; they might have been able to inform researchers whether future research on their samples and data was acceptable.

   b. Researchers would have gained a better understanding of potential research they might need to explicitly discuss during the consent process.

2. **What are some ways in which researchers might have engaged with the Havasupai community?**

   Starting points for discussion:

a. Informational/educational meetings to educate potential participants about the research process and goals.

    i. Opportunities for potential participants to voice their reasons and goals for participating.
    ii. Opportunities for researchers to offer clear explanation of what it means to donate a biological sample—and to discuss what will happen to the samples following the research project.
    iii. Ability to consider varying levels of education in drafting informed consent; researchers must ensure that language in informational material and consent documents is composed at a comprehensive level appropriate for the participating population.

Scenario B. *Data on breast cancer in African American women—including genetic data from family linkage studies, and survey and recruitment data related to breast cancer testing and prevention behaviors—are scarce, despite the fact that African Americans are more likely to be diagnosed with the condition and less likely to survive it.*[7] *In an attempt to address this disparity, genetic researchers partnered with a community organization to gain insight from members of the population, in order to increase participation and aid recruitment for research. Although this particular example illustrates research using a genetic test rather than whole genome sequencing—that is, a specific targeted test rather than a survey of the entire genome—many of the same principles apply.*

Below is a brief summary of the study; you can read more here:

> Ochs-Balcom, H.M., Rodriguez, E.M., and D.O. Erwin. (2011). Establishing a community partnership to optimize recruitment of African American pedigrees for a genetic epidemiology study. *Journal of Community Genetics*, 2(4), 223-231.

In order to optimize recruitment for a familial breast cancer genetics study in an African American population, researchers partnered with the National Witness Project, a community-based breast and cervical cancer education project. The team convened focus groups of African American women and ascertained some of the challenges of recruiting this population into breast cancer genetic studies. Themes identified through the focus

[7] National Cancer Institute. (2008). *Cancer Health Disparities*. Retrieved from http://www.cancer.gov/cancertopics/factsheet/disparities/cancer-health-disparities; Hughes, C. et al. (2004). Minority recruitment in hereditary breast cancer research. *Cancer Epidemiology, Biomarkers & Prevention*, 13(7), 1146-1155; Ochs-Balcom, H.M., Rodriguez, E.M., and D.O. Erwin. (2011). Establishing a community partnership to optimize recruitment of African American pedigrees for a genetic epidemiology study. *Journal of Community Genetics*, 2(4), 223-231.

groups included communication barriers and shame and stigma associated with a cancer diagnosis.

Researchers felt strongly that because minority communities have poorer health outcomes following a cancer diagnosis, community engagement is especially important in studies with these communities. Better understanding the barriers to study participation can aid in recruitment and fill holes in knowledge and data, which can ultimately improve health outcomes. After gathering community input from the focus groups, the researchers altered recruitment strategies in response to the demonstrated concerns. At the time of publication of the paper, the researchers were midway through the recruitment process, and were finding it to be successful.

1. **What distinct recruitment challenges came up in this study that might also arise in similar types of research? How might community engagement address those challenges?**

   Starting points for discussion:

   a. The authors of the article describe the unique concerns raised by this minority population, including a lack of knowledge about genetics, the shame or stigma experienced in association with a cancer diagnosis, and heightened privacy concerns as compared with other populations.

   b. Community engagement might address these challenges by providing the participants greater access to the researchers, building more transparency into the study design, carefully assessing attitudes towards privacy and confidentiality, and connecting the community with other resources and support.

2. **In general, how can community engagement in genetic and genomic research address and potentially ameliorate health disparities? How can community engagement help frame and address issues with recruitment, study design, and publication of results?**

   Starting points for discussion:

   a. As mentioned in the article, lack of engagement with members of the African American community in research has been considered a crucial factor in current disparities in the effectiveness and progress of health in the United States.

b. There are few studies of African American women concerning family linkage and breast cancer, causing many African American women to undergo suboptimal treatments.

c. Through engaging members of minority communities in genetic and genomic research, researchers might gain more opportunities to study disease in minority populations and therefore determine the most effective therapies for these patients.

3. **This breast cancer study dealt specifically with community engagement to enhance recruitment. What other strategies for engaging the community might be helpful in the later stages of research? What benefits do these strategies provide?**

   Starting points for discussion:

   a. Potential answers include: involving the community in planning the study design, conducting follow-up studies, developing informed consent procedures, and planning of data analysis and interpretation; and developing plans for post-publication dissemination of results back to the community.

Scenario C. *Biobank research presents its own ethical challenges and involves a set of circumstances that make community engagement more difficult but no less important. In the case summarized below, researchers describe the nature and difficulties of tailoring aspects of biobanking to individual and community preferences.*

Below is a brief summary of the study; you can read more about it here:

> Gottweis, H., and G. Lauss. (2012). Biobank governance: Heterogeneous modes of ordering and democratization. *Journal of Community Genetics*, 3(2), 61-72.

This article describes the heterogeneity inherent in biobanks. As a result, the authors suggest that biobank governance must be flexible, and that managers must gather the input of the many interrelated groups of stakeholders in order to develop sound policy. They go on to describe various biobanks and the methods of governance for each. They conclude that biobanks cannot be separated from the bodies of the tissue donors, and, as a result, cannot be divorced of their social and political implications. Engaging the public in order to ascertain their views and maintain their trust is an important part of

biobank governance, the authors explain. But discerning who constitutes "the public" for any particular biobank can be a challenge in itself.

1. **Who is the "community" in biobank research, and why is it difficult to determine who should be included in community engagement efforts?**

   Starting points for discussion:

   a. To read more about complications in biobank research, see *Privacy and Progress*, p. 54.

   b. There are many ways in which "community" could be defined in this instance: prospective patients in a clinical setting whose biological samples might be stored in a biobank, local community members living near a research hospital, or members of local professional organizations (e.g., nurses, doctors, physician assistants, laboratory technicians). Consider the challenges of engaging any of these particular communities.

   c. However community is defined for the purposes of engagement in biobank research, representatives of the community should be consulted on research design options including how to select biobank samples for research (e.g., at random, based on disease status, carrier status).

2. **What are some ways that biobanks can differ from each other, and how does this variation affect the ways in which community engagement is deployed?**

   Starting points for discussion:

   a. Biobanks can have very different groups of individuals whose biological samples and data are included. For example:

      i. Data can be collected from patients in a clinical setting or participants in a research setting;
      ii. Data might be identifiable, de-identified, or anonymized;
      iii. Data can be disease-specific (e.g., from patients with breast cancer), or data can come from healthy individuals; and
      iv. Data can represent individuals from one community, nationality, or race, or from many.

   b. Community engagement can be deployed regardless of the identity of the community. The community sought might be broader in some cases (e.g., a group of healthy individuals from varied backgrounds who participated

in a sleep study) than others (e.g., breast cancer patients) and the ways that researchers reach out to those communities should be tailored to each specific group.

Scenario D. *The HapMap Project is a large-scale genomic research project that attempts to draw comparisons among various populations of the world. DNA samples are taken from ethnically homogenous groups of people from disparate locations around the globe. The research has the potential to teach us about our commonalities and describe our diversity. But some might find it ethically troubling because of the potential to enhance racist attitudes or to lead to stigmatization of certain populations For these reasons, the HapMap Consortium has made significant efforts to engage communities in the research process.*

Below is a brief summary of the project. You can read more about the HapMap Project and community engagement here:

Rotimi, C., et al. (2007). Community engagement and informed consent in the International HapMap Project. *Community Genetics*, 10(3), 186-198.

The HapMap Project is an effort to identify commonalities and differences among the genotypes of human populations around the world. The HapMap researchers recruited populations from Nigeria, Japan, China, and the United States. They understood the complex ethical implications of their research—even though they obtained informed consent from all participating individuals, they knew that results of the study were likely to implicate entire populations of people, not just the participants themselves. As a result, they made an effort to collect community input and to alter study design in response to concerns.

Researchers identified four important goals for community engagement: 1) to ascertain views about the ethical and social implications of the study, both for study participants and for populations in general; 2) to gather input from each population as to preferred methods of sample collection and preferred description of the population; 3) to convey extensive information about the project so that participants were fully informed before deciding to participate; and 4) to develop a line of communication in order to keep participants informed of future developments.

Although some parts of the scientific protocol could not be altered, the researchers attempted to use community input to alter the project wherever appropriate and possible. The researchers believed that in conducting such sensitive research a spirit of openness and transparency was absolutely essential. The researchers described the community engagement process positively, overall. However, they noted one limitation of their

efforts. They only sought input from the communities that were a part of the project and expressed regret that they were not able to ascertain how the research might affect other populations, such as minority communities in the countries where the research was conducted.

1. **HapMap researchers admitted that there was a flaw in their community engagement protocol. What do they think they could have done differently, and how might it have affected the outcome and experience?**

   Starting points for discussion:

   a. See page 194 of the article. Researchers admitted that they might have gathered more critical input if they had engaged minority communities that were not included in the research. Those communities might have expressed concern about being excluded. And other communities that were not included might have expressed concern about the research itself, especially if conclusions to be drawn might ultimately be compared to and attributed to other similar groups.

2. **What are the differences between using community feedback to shape a scientific protocol and using community engagement to change pre- and post-study practices, such as recruitment and data reporting? Where do the HapMap researchers think this line should be drawn?**

   Starting points for discussion:

   a. On page 194 of the article, the researchers describe their process for seeking community engagement in labeling and identification of samples. Some aspects of labeling could not be changed even after community input due to practical purposes. Particularly, the authors note that the samples needed to be labeled CEPH, the name of the study, in order to avoid confusion in the scientific community. To what extent should practical or logistical reasons for a particular aspect of protocol take priority over the interests of a community?

3. **When genetic research of a racial or ethnic and potentially sensitive nature is being conducted why is community engagement particularly important?**

   Starting points for discussion:

a. See page 189 for an explanation of the researchers' goals regarding community engagement in this type of research. They include:

    i. Ascertaining views about the ethical and social implications of the study, both for study participants and for populations in general;
    ii. Gathering input from each population as to preferred methods of sample collection and preferred description of the population;
    iii. Conveying extensive information about the project so that participants were fully informed before deciding to participate; and
    iv. Developing lines of communication in order to keep participants informed of future developments.

4. **In this example, what information might have been omitted from the community engagement process, and how might researchers make up for it?**

   Starting points for discussion:

   a. As the article explains, minority communities and others were not included in study design and therefore were not given the opportunity to provide input.

   b. Consider how researchers should use the principles of responsible stewardship and justice and fairness (described in detail in the Community Engagement Background module) to ensure that even when certain communities are not engaged their interests can be taken into account.

VII. Exercises

Exercise A. *In light of the Bioethics Commission's recommendations, especially Recommendation 1.1, which can be found reprinted on the first page of this module, think critically about how best to use community engagement to reconcile the need to advance whole genome sequencing technologies for the public good with the requirement to protect the individual privacy interests of those who share their whole genome sequence data for the benefit of research. Consider how one might use the valuable insight of the community to promote fair use and sharing of genomic data.*

1. **While considering the complicated questions about informed consent in the context of whole genome sequencing, outlined by the Bioethics Commission (on pages 87-91 of *Privacy and Progress*), how can input from the community enhance or supplement the informed consent process?**

a. Consider whether there could be special needs in certain communities that would make input from their members important in revising the informed consent process (e.g., language, education level, and number and type of vulnerable members).

Exercise B. *In her presentation to the Bioethics Commission in August 2012, Dr. Laura Lyman Rodriguez of the National Human Genome Research Institute noted that the research community needs to understand that "there are patient-driven research objectives now," and that individuals are "coming together to do [research]."*[8] *Transcripts and archived video of Dr. Rodriguez's presentation are available on the Bioethics Commission's website under Meeting 10, Session 4 (beginning at 10:36 on the webcast video).*

Investigate the meaning of "patient-driven research," (for an example, see the following website: http://www.personalgenomes.org).

a. How does patient-driven research differ from community engagement in traditional research? What is the role of the researcher in each? Can you learn any lessons from patient-driven genomic research that might help enhance community engagement in your research or area of expertise?

VIII. Glossary of Terms

Anonymized data: Data from which personal identifiers have been permanently removed and no link to the individual remains.

De-identified data: Data that have been separated from information identifying the individual from which they were derived. A "key" or code connecting the two might still exist, but recipients of the data are not allowed to access the key.

Democratic deliberation: A method of decision making to address an open policy question in which participants consider both relevant information and ethical aspects, justify their arguments with reasons, and treat one another with mutual respect, with the goal of reaching an actionable decision for policy or law, open to future challenge or revision.

[8] Rodriguez, L.L., Director, Office of Policy, Communications, and Education, National Human Genome Research Institute. (2012). Roundtable Discussion. Presentation to the Presidential Commission for the Study of Bioethical Issues (PCSBI), August 1. Retrieved from http://bioethics.gov/node/741.

Distributive justice: The ethical principle that calls for equitable distribution of benefits and burdens across society—for example, the benefits and burdens of biomedical research, or of technological advances.

Informed consent: The process of informing and obtaining permission from an individual before conducting medical or research procedures or tests.

Intellectual freedom and responsibility: The notion that scientists and researchers, acting responsibly, should use their creative abilities to advance science and the public good while adhering to the ideals of research, avoiding harm to others, and abiding by all associated rules and regulations.

Public beneficence: The ethical principle that calls on researchers, scientists, and decision-makers to pursue and secure public benefits while minimizing personal and public harm.

Respect for persons: The ethical principle that calls on health professionals and researchers to treat individuals as independent and self-determining (autonomous) agents and to provide additional protections to persons with diminished autonomy in clinical care and research settings.

Responsible stewardship: The ethical principle that calls on governments and societies to proceed prudently in promoting scientific advancement by taking into account the interests and needs of individuals who may not be in a position to represent themselves.

Whole genome sequencing: Determining the order of nucleotide bases—As, Ts, Gs, and Cs—in an individual's or organism's entire DNA sequence.

IX. Additional Resources

Cornel, M.C., van El, C.G., and W.J. Dondorp. (2012). The promises of genomic screening: Building a governance infrastructure. Special Issue: Genetics and Democracy. *Journal of Community Genetics*, 3(2), 73-77.

Gutmann, A. (2013). Data re-identification: Prioritize privacy. *Science*, 339(6123), 1032.

Gymrek, M., et al. (2013). Identifying personal genomes by surname inference. *Science*, 339(6117), 321-324.

Hedlund, M., Hagen, N., and U. Kristoffersson. (2012). Genetics and democracy. *Journal of Community Genetics*, 3(2), 57-59.

Juengst, E.T. (2003). Community Engagement in Genetic Research: The "Slow Code" of Research Ethics? In Knoppers, B. (Ed.). *Populations and Genetics: Legal and Socio-Ethical Perspectives*. Herndon, VA: Brill Academic Publishers, pp.181-198.

Mello, M.M., and L.E. Wolf. (2010). The Havasupai Indian trip case—Lessons for research involving stored biologic samples. *New England Journal of Medicine*, 363(3), 204-207.

Terry, S.F., et al. (2011). Community engagement about genetic variation research. *Population Health Management*, 15(2), 78-89.