



ASIP Public Affairs

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If You Use Human Tissue in Your Research, You Should Know About ELSI

by Dr. Mark Sobel

In the early 1970s, scientists at the forefront of the new field of DNA recombinant technology voluntarily put a moratorium on their research to prospectively consider the societal ramifications of their endeavor. The result was an immensely successful plan, carefully constructed by the scientists themselves, to set safety standards. Research proceeded in a safe, effective manner without burdensome regulations.

Given this precedent, when the **Human Genome Project** was originally funded by the US Congress, a percentage of funds was earmarked to prospectively consider the ramifications of this incredible undertaking. The **Ethical, Legal and Social Implications (ELSI)** Program of the National Center for Human Genome Research was thus established. The ELSI Working Group is a body of geneticists, ethicists, attorneys, social workers, patient advocates and public health workers responsible for developing ethical studies and oversight. Recent initiatives by the ELSI Working Group have included workshops, task forces to develop policy and the funding of multicenter pilot genetic screening programs.

The rapid pace of advances in human molecular genetics has created new questions in medical ethics. Given a society that values individual privacy and that does not provide universal health coverage (with the very real possibility that health insurance can be denied for pre-existing conditions such as a genetic predisposition), it was inevitable and necessary that the issue of genetic privacy and the protection of patient's rights be considered. Recently, the ELSI Working Group has been considering a policy initiative on informed consent for genetic research, including research on stored tissue samples. In a technological age in which DNA can be extracted from minuscule archival samples, including frozen blood or tissue samples such as urine, sputum, and even paraffin-embedded tissue blocks, genetic tests can be potentially performed on virtually any stored tissue sample.

Currently, retrospective research on anonymous tissue samples is exempt from review by institutional review boards (IRBs). Thus, as long as the tissue sample was originally obtained for clinical testing purposes and has been stripped of patient identifiers to protect patient confidentiality, it can be used for any research or for clinical control purposes. Many members of the ELSI Working Group, however, have expressed concern that

(1) many patients are not adequately informed that some of their tissue or blood (that has been removed for performing a specific test or that was surgically resected) is stored indefinitely and may eventually be included in a retrospective research study;

(2) many 'anonymous' samples can be theoretically traced back to the patient's hospital chart, using coded research sample numbers;



(3) genetic testing results on such samples can have adverse psychological and socioeconomic consequences for the patient as well as for members of the extended family;

(4) patients have an essential right to be kept informed, on a periodic basis, of the progress of research studies in which their samples are being used, including release of information prior to publication of results in the medical literature.

The ELSI Working Group has therefore proposed for consideration that all proposals for anonymous research on previously stored tissue samples as well as on samples to be collected in the future should be reviewed by an IRB and that individuals from whom tissue samples are collected should be given the option of refusing to have research undertaken. Thus, if a researcher wished to use a previously stored sample for any purpose, the patient who originally provided the sample would have to be contacted and informed of the specific test to be performed. If deceased, the executor of the estate and/or other surviving relatives would have to provide consent. Furthermore, it has been recommended that for newly collected samples, patients be given a consent form with options that would include

- (1) whether any research at all can be conducted,
- (2) whether some specific research can be conducted, but with the option of excluding research on particular diseases,
- (3) whether name identifiers should be included or not,
- (4) whether samples can be shared with other researchers,
- (5) whether the patient wants to be contacted about the findings of the study,
- (6) whether they wish to be informed of potential profits derived from the research,
- (7) the option of removing the sample from the study at any time.

A draft of a 'Genetic Privacy Act' has been funded by ELSI and many state legislatures are using this draft as a model to pass laws that would regulate the use of tissues samples. As these new proposed policies have begun to be discussed many individuals in the pathology community have expressed concern that pathologists, who traditionally have been responsible for keeping and safeguarding the integrity of stored tissue samples, have not been adequately consulted during the ELSI Working Group discussions. How would such a program of informed consent be administered? Many researchers are worried that the new ELSI initiative would impede or prevent most ongoing human molecular research studies as well as any other studies that do not involve genetic research.

How shall we balance the rights of the patient with the need for our society to advance research on human disease to provide better health care? Currently under review by many professional societies interested in research is the following text that was recommended by the Molecular Pathology Resource Committee and the Commission on Anatomic Pathology of the College of American Pathologists: While we agree that the principles of utilization of stored tissues must continue to ensure patient privacy and confidentiality, we emphasize that any guidelines on the ethical, legal and social impact of such use must also preserve and protect the accessibility of residual human material for the development of new diagnostic and therapeutic modalities for human diseases.

What is your position and how would you like the ASIP to respond to this issue?



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