ACP
American College of Physicians
Internal Medicine | Doctors for Adults

David L. Bronson, MD, FACP
President

Professor of Medicine, Lerner College of Medicine of
Case Western Reserve University
President and CEO, Cleveland Clinic Regional Hospitals
Executive Board Office – JJN6-01
9500 Euclid Avenue
Cleveland, OH 44195

Phone: 216 445-2740, Fax: 216 445-2778
Email: bronsod@ccf.org

May 24, 2012

Amy Gutmann, PhD, Chair
Presidential Commission for the Study of Bioethical Issues
1425 New York Avenue, NW
Suite C-100
Washington, DC 20005

Dear Dr. Gutmann:

The American College of Physicians (ACP) submits this letter in response to the Commission's request for comments regarding the ethical implications of evolving notions of privacy and access in relation to the integration of large-scale human genome sequencing into research and clinical care. The American College of Physicians is the largest physicians' specialty society and second-largest physician membership organization in the United States. ACP represents 132,000 internal medicine physicians and medical student members. Internists specialize in primary and comprehensive care of adolescents and adults.

The Commission is seeking comments regarding the implications of large-scale human genome sequencing for the privacy of individuals, research subjects, patients and their families. The Commission is interested in receiving views on evolving notions of privacy as evidenced and influenced by social media and on models and mechanisms for protecting the confidentiality of genetic/genomic and other sensitive information stored in databases and biobanks. In addition, the Commission has invited interested parties to comment on the related issues of balancing individual and societal interests regarding the sharing of and access to large-scale human genomic sequencing data.

Issues of privacy and confidentiality related to the storage and use of genetic materials have implications for both clinical medicine and medical research. In the clinical context, maintaining the privacy and confidentiality of the patient's information reflects respect for the patient and contributes to trust in the physician.[1] The patient-doctor relationship is dependent upon such trust, which must extend to the confidentiality of personal information shared as part of that relationship. In order for patients to be willing to disclose to their physicians all information that is pertinent to their care and treatment, they must feel secure that such information will not be inappropriately disclosed.

"Confidentiality is a fundamental tenet of medical care."[1] Changes in medical practice, including the introduction of new electronic storage and processing of health records, the use of e-mail and other electronic means of communication, and third-party payment for medical services have made maintaining a patient's privacy and the confidentiality of health care

information more challenging. Physicians need to be scrupulous in following appropriate security protocols for the storage and transmittal of information and in adhering to best practices for electronic communication and the use of decision-making tools. A patient's personal health information should not be released without the patient's consent.[1]

Genetic testing may present the physician with complex ethical problems, such as whether a particular family member or members should be informed of the test results. "Although information about the presence of a genetic risk factor or genetic disease in a family member raises the possibility that genetically related individuals are at risk,"[1] the physician's primary obligation continues to be to the patient and to his or her best interests. The potential implications for family members should be discussed with the patient prior to genetic testing and included in the consent process. Based upon testing results, the physician should encourage the patient to contact family members who may be at risk or, alternatively, should obtain the patient's consent to contact potentially affected family members about test results and genetic counseling.

The move toward electronically recorded and stored health information over the past decade has led to increased concern over the protection of patient privacy and the confidentiality of personal health information. The accumulation of genetic information and the greater availability of genetic data through electronic storage heighten both the concern over and the potential impact of privacy breaches. Inadvertent or unauthorized disclosure of Information obtained through genetic testing may significantly affect not only the life of individual patient whose biological material is being used but also members of his or her extended family. Identification of factors that may be perceived as creating a "genetic predisposition" to specific diseases or conditions in the subject as well as in his or her family members can have implications for critical decisions such as marriage and child-bearing. Disclosed genetic information may be used in contexts other than health care including employment, insurance, and law enforcement and may lead to discrimination and other serious harm.[1]

Regarding research, ACP recognizes the importance of balancing patients' needs and preferences for genetic information to remain confidential and secure with researchers' need for access to complete, accurate and available health information. However, "[f]or many individuals there is no difference between health care providers and researchers, especially when the providers and researchers work for the same institution and patient-based clinical records are used in the research." [2] As a result, a loss of confidence in researchers' commitment or ability to maintain the confidentiality of genetic information will spill over to clinicians and other health care providers. With such loss of confidence, patients may withhold essential information from their physicians with resulting negative clinical consequences for themselves as well as for others. [1]

ACP believes that all attempts should be made to de-identify biological materials.[3] However, we recognize the reality that biospecimens may not be truly de-identifiable and that even when de-identified, biological materials are not anonymous. As stated in the advance notice of proposed rulemaking related to proposed changes in the Common Rule, ". . .what constitutes "identifiable" and "de-identified" data is fluid; rapidly evolving advances in technology coupled with increasing volume of data readily available may soon allow identification of an individual from data that is currently considered de-identified. In this sense, much of what is currently considered de-identified is also potentially identifiable data." [4] In the absence of means of assuring permanent de-identification of genetic information, ACP asserts

that there should be tighter controls against improper re-identification of de-identified patient or subject data.[3]

In light of the very significant impact that inappropriate disclosure of research subjects' genetic data may have, ACP believes that researchers should be required to obtain fully informed and transparent consent from the subjects of genetic research.[1] Fully informed and transparent consent requires the disclosure of all potential uses of patient biological materials and data. The consent process should be in language understandable to research subjects and needs to include the preference of the research subject regarding future contact for notification about results and/or consent for additional research participation. Research should be limited to the use specified by the protocol during the informed consent process. Research subjects should be made aware that it may not be possible to withdraw their biological materials from research use once they are de-identified. Full disclosure of the risks and benefits of research involving biological material allows research subjects to make well-informed decisions.[1]

Consent is also an issue of concern with respect to the use of existing genetic materials. The ACP recognizes that additional study is needed to address issues of informed consent regarding the use of existing biospecimens and genetic data in future research. Several models of informed consent in this context have been suggested, including specific consent, i.e., reconsenting by the subject for the new use of his or her biospecimens or genetic data; tiered or graduated requirements for consent; general or open-ended consent to all future uses of genetic materials and information subject to IRB review; and blanket consent with no restrictions on future use. The Institute of Medicine included recommendations for use of existing biological materials and/or genetic information in its 2009 report on privacy and research[5]. The IOM has recommended two conditions for the use of such materials and/or information in future research: First, that the original consent for the use of a subject's biological materials and/or genetic information describes the types or categories of research that may be conducted using the materials and/or information and, second, that "an IRB determines that the proposed new research is not incompatible with the initial consent and authorization and poses no more than a minimal risk."[5] In light of the significant impact that disclosure of subjects' genetic data may have, ACP does not support proposals that would excuse research protocols conducting research with previously collected biospecimens and/or genetic information from continuing IRB review.[4]

Evolving technologies and scientific advances challenge but do not change the ethical obligations of physicians and other health care providers. Trust is essential to the physician-patient relationship and also to public support for ongoing and future medical and genomic research. Maintaining that trust requires that patients and research subjects be assured that genetic information derived from their biological materials will be kept confidential and that they be made aware of the intended uses of such materials and have the opportunity to consent, or not consent, to such uses. We believe that these challenging issues warrant significant discussion and exploration among a broadly representative array of stakeholder groups.

Sincerely,

[1] Snyder L. American College of Physicians Ethics, Professionalism, and Human Rights Committee. Ethics Manual, sixth edition. Ann Intern Med. 2012; 156:73-104.
[2] Rothstein MA. Is deidentification sufficient to protect health privacy in research? Am J. Bioeth. 2010; 10:3-11.
[3] American College of Physicians. Health Information, Technology and Privacy: A Position Paper. July 2011
[4] Human subjects research protections: enhancing protections for research subjects and reducing burden, delay and ambiguity for investigators. 76 Federal Register 143 (July 26, 2011) pp. 44512-44531.
[5] Institute of Medicine. Beyond the HIPAA privacy rule: enhancing privacy, improving health through research. Washington, DC: National Academies Press; 2009.