



26 July 2006

Secretary's Advisory Committee on Genetics, Health and Society
ATTENTION: Ms. Amita Mehrotra
NIH Office of Biotechnology Activities
6705 Rockledge Drive, Suite 750
Bethesda, MD 20892

Dear Ms. Mehrotra:

Thank you for the opportunity to comment on the Committee's May 2006 *Draft Report on Policy Issues Associated with Undertaking a Large U.S. Population Cohort Project on Genes, Environment, and Disease*. The IEEE-USA strongly believes that electronically accessible healthcare information, including genetic and environmental data, offers the possibility of improving the quality of life of all our citizens, while, over the long term, potentially reducing the costs of our health care delivery system by improving population health and re-focusing our current system into disease prevention, rather than diagnosing, treating and curing disease.

IEEE-USA recommends that the following aspects of the report should be modified to provide the Secretary the best range of policy options for implementing this national resource:

- **Policy Context:** Our primary concern is that the report envisions this effort as a "project," rather than as a component of the larger effort to "... *make wider use of electronic records and other health information technology, to help control costs and reduce dangerous medical errors*" as President Bush directed in his State of the Union address. To fulfill the President's goal, we believe that all possible environmental and healthcare information of our nation should be put to use for the benefit of our citizens.
- **Privacy:** Privacy considerations should not be ignored or minimized, but should be viewed as a constraint, not as a roadblock.
- **Genetic Sample Selection:** We are concerned about the approach the report takes to the issue of acquiring a representative sample of genetic information. Race and ethnicity are very limited and imprecise tools to use in achieving such a sample.
- **Policy Issue Resolution:** There is a tacit assumption that few people would voluntarily participate unless most or all policy issues are resolved in their entirety. With appropriately informed consent, adequate participation may well be achieved while many issues remain open.
- **Intellectual Property:** We believe the issues of intellectual property rights must be included in the policy analysis in more depth, not just for the genomic data collected, but also for derivative products based on that data.

- **Other Agency Involvement:** Finally, we believe that not only the National Institute of Health, but all the Agencies within the Department of Health and Human Services as well as other Federal Departments, need to integrate their research knowledge, including that gained from the prospective project.

These concerns are described in more detail in the attachment. Thank you again for the opportunity to comment on the report.

IEEE-USA is an organizational unit of The Institute of Electrical and Electronics Engineers, Inc., created in 1973 to advance the public good, while promoting the careers and public-policy interests of the more than 220,000 engineers, scientists and allied professionals who are U.S. members of the IEEE. If you have any questions or we can be of further assistance, please contact Deborah Rudolph in our Washington DC office at (202) 530-8332; d.rudolph@ieee.org.

Sincerely,

A handwritten signature in black ink that reads "Ralph W. Wyndrum, Jr." in a cursive script.

Ralph W. Wyndrum, Jr., Eng.Sc.D.
President, IEEE-USA

RWW/dr:bc

Encl.

IEEE-USA Comments on the Public Comment Draft “Policy Issues Associated with Undertaking a Large U.S. Population Cohort Project on Genes, Environment, and Disease”

Policy Context - Stand-alone Project or an Element of Electronic Health Record (EHR) Implementation

A research effort that combines genetic, environmental, and healthcare information will require a wide range of data from which to draw conclusions. For many research purposes, it is the combination of an individual's genetic background with the same individual's environmental and healthcare experience that provides the information necessary to draw conclusions that may result in improved care. Diagnostic, treatment, and outcome data constitute a minimum set of information required for research to meet the President's goals for healthcare. To reduce the uncertainty of research conclusions, a lifetime longitudinal EHR (including environmental and genomic data) would be ideal.

In essence, the Population Cohort Project would add a genomic (and perhaps environmental) component to the EHR. Incorporating genomic data and research into the EHR model simplifies and clarifies the issues of public engagement and project value. The addition of these types of information to an EHR should be part of the public engagement process for EHRs.

A continuing goal of the research process should be to determine what the addition of genomic and environmental information does to improve outcomes over research that does not include such information. The recent example of the variability of the impact of some beta-blockers on heart disease patients with different genetic makeup shows how genomic information can potentially decrease costs by targeting the most effective drugs to specific patient populations. More generally, the cost and treatment benefits of adding genetic and environmental information to other EHR data should be the focus of the genetic and environmental aspects of the overall EHR effort. This approach to thinking about and discussing the genetic and environmental data components should help with the process of public acceptance when compared to discussing a stand-alone, large-scale genetic project in the abstract. We believe the establishment of a large genetic data base is best treated as an element of providing the maximum benefit of EHRs. A stand-alone genetic data base is not the ultimate goal, and would likely not gain public support. At the same time, there is no need to wait for the availability of genomic information in order to produce useful EHRs. While the conceptual approach to incorporating genomic data into EHRs should be developed now, actual implementation of a substantial genomic data component may need to wait for the cost of obtaining the data to drop.

Treating a genomic data base as a separate project, rather than as an integral part of EHR implementation, would also cloud the cost aspect of including genomic data in research. Today, individuals can get limited genetic information via commercial providers for a relatively small fee. Such information could immediately be added to the individual's Personal EHR. On the other hand, Professor George Church at Harvard Medical estimates that obtaining a complete individual genome currently costs around \$20 million. He expects that the cost will reduce to \$20,000 by around 2010, and a cost of \$1000 might be expected by 2014 (these estimates may not include DNA outside of the human genome, some of which may turn out to be of medical interest). Given the expected rapid decline in cost, adding genetic information incrementally may be the best policy (a sort of “dollar cost averaging” approach). This would seem to be most easily done in the context of the larger EHR effort. It would allow researchers to use what they have, when they get it. In this context, genetic information obtained as a result of the National Children's Study might constitute a significant initial element of genetic information.

Privacy Considerations

HIPAA provides the basic framework for healthcare privacy, and, within that framework, we believe research activity should be able to operate successfully. While many individuals may feel strongly that access to their medical information should be limited to only their medical providers, many others are likely to be willing to share their EHRs, including genetic information, for research purposes. Individuals may be willing to share that information as de-identified information, as information with a decoupled back-link, or as fully identified information for research purposes only. The implications of each option should be fully explained as part of the public engagement process, within the context of an EHR and other medical information. It should be noted that de-identified data has very serious drawbacks since it makes it difficult to maintain the longitudinal data across multiple providers and could produce duplicate records that could not be identified as such.

With properly informed consent, it seems highly likely that an adequate pool of volunteers can be acquired to begin populating a genomic data base, integrated with other EHR data. While most people don't donate organs, give blood, or participate in clinical trials, a significant number do. There seems to be no reason for greater pessimism with respect to providing genomic information. The current Behavioral Risk Factor Surveillance Surveys provide a model for the collection of individually identified data in the environmental area that could be adapted to genomic data in the near term. A likely pool of volunteers is those individuals that are assured of lifetime medical care. This includes military and civil service retirees and individuals under Medicare. While these groups do not include all age ranges, they could serve as a test for the adequacy and evolution of technical privacy measures to gain confidence before moving to a general population. The National Children's Study could be used to develop techniques for managing information at the other end of the age spectrum.

Genetic Sample Selection

If the project goal is to acquire a sufficiently varied genomic sample to draw scientific conclusions about the role of genes and the environment on disease, then "Recruitment of Racial/Ethnic Groups" may be an inadequate approach to acquiring diversity. People are allowed to "self-identify" their ethnicity/race in census information. A "Latino" might be a Catalonian from Spain, a mixed-blood Mestizo from Mexico, or a person of African descent from the Dominican Republic. The whole issue of identifying population subgroups is politically sensitive and would create its own difficulties. It appears that these difficulties have already appeared, since two versions of the report that have appeared for review have significant changes in this area.

Several concerns need to be addressed in acquiring genomic information that will be of benefit to healthcare. From a scientific perspective, at least two goals may need to be met in choosing participants.

One scientific goal is to acquire genetic information that is likely to lead to the identity of genes that play a role in disease. This goal may be best met by assuring that the sample contains a significant number of individuals with a specific disorder that can be compared to a sample without it, ideally under a variety of environmental conditions. To meet this goal, some recruitment should occur in a population that has the disorder. For example, Americans with the historic geographic origin of sub-Saharan Africa are more likely to suffer from sickle cell anemia than most European Americans. This is not because of their "race", but because the same genetics that predispose them to sickle cell also protect against malaria, a genetic trait shared with many Mediterranean peoples, including Arabs and Italians. Enrollment of entire families, across multiple generations, where there is a known risk of a disease, is another approach to meeting the goal of including groups at risk of a disease.

A second scientific goal is to acquire sufficient genetic diversity, together with environmental information, to perform broader “data mining” analyses of diseases, adverse drug reactions, etc. This goal is probably best met via a random sample across U.S. census districts chosen to vary environmental conditions.

To meet either of these goals, race and ethnicity are probably not the key elements of information that should tag individual genomes in the data. The more appropriate tags may be “ancestral origin, heritage, history and environmental exposure” (Report, p.33) or similar data items that might serve to group individuals with a propensity for a specific disorder. For some interim (possibly quite long) period, this sort of information could be used to allow individuals with an unknown genome to determine their level of risk so that they can take precautionary measures (if any are available). If we get to the point in the future that one’s personal genome is part of one’s personal EHR, then these background factors become less important, since a direct determination can be made of whether the specific genetic and environmental risk factors are present for the individual. As the sickle cell anemia example suggests, this information would be at a much finer granularity than the concepts of race and ethnicity would permit.

Finally, there are political goals that must be met as well, in terms of perceived fairness in including the genetic information of (perhaps self-identified) racial and ethnic groups and of socio-economic groups. One approach to insuring that fairness is to collect self-identified information on these factors in the process of identifying individuals that meet the scientific goals above. Information on the distribution of these factors can then be evaluated by an appropriately diverse oversight group to determine if there are obvious fairness shortcomings, and additional individuals could be recruited to provide a more balanced population if required.

At least initially, a further limitation on participation may be concerns about the impact of even determining an individual’s genetic information. Persons identified as potential participants by healthcare providers might be reluctant to participate in such studies, fearing that this might have an adverse effect on their ability to obtain healthcare or healthcare insurance in the future. This is a realistic fear, given that we still do not have adequate legal protection against genetic discrimination (p. 45). There is also the ethical dilemma posed (p. 38) by having the same person assume the dual roles of healthcare recipient and research subject. These concerns may cause a further limitation of at least the initial pool to groups, such as those mentioned under privacy considerations above, that have guaranteed access to healthcare.

Relationship of Policy Issue Resolution and Project Initiation

Efforts to acquire and use genomic data, such as the Personal Genome Project and commercial gene tests, are already underway. As we have suggested, there are likely to be a significant number of volunteers to participate even with multiple policy issues undecided. As the policy issues are resolved, additional participants are likely to be available. Unresolved policy issues are therefore unlikely to be a roadblock to proceeding with the integration of genomic data into EHRs in ways that are compatible with healthcare research. A larger issue is the lack of “first mover” research using an EHR (with or without genomic information). Current research activities tend to develop their data structures and data bases on an individual case basis, which limits the utility of the information collected for other research activities. The National Children’s Study is well along in developing the framework necessary to make use of EHRs to support a range of research activities. It, and other research efforts, should begin the planning for the use of EHRs conforming to standards. The IEEE-USA white paper “Interoperability for the National Health Information Network” provides background on what would be required. The Certification Commission for Healthcare Information Technology (CCHIT), within the Office of the National Coordinator for Health Information Technology, has prepared guidelines for certification for EHRs for Ambulatory Care along the lines suggested in the IEEE-USA white paper. A joint effort between research elements of the Department of Health and Human Services and CCHIT to provide certification criteria for healthcare research applications (including genomic data) could speed the process of creating interoperable research data that could be used in a broad range of research activities.

Intellectual Property Rights

We agree with SACGHS that intellectual property (IP) policy is critically important and needs to be clarified at the outset of the project, and we agree that the scale of the project may necessitate public-private partnerships in order to mobilize adequate funding and human resources. However, the Draft Report has not adequately emphasized the close interrelationship between these two issues: Whether there are incentives for private funding and participation will depend, rather critically, on the question of data access and control over discoveries that may result from research with the data and tissues collected in the study. Treating IP policy and public-private partnerships as two separate policy issues may result in sub-optimal choices that fail, in the end, to achieve maximal public benefit from the proposed database and tissue banking efforts. Therefore, IEEE-USA believes that it is critical to consider these two issues together, with an emphasis on the following points:

- This project actually involves at least three separate phases and objectives: (1) creation and development of a national database and tissue resource, (2) secondary use of that resource in research, and (3) successful translation of resulting discoveries into medical products and treatment modalities to improve clinical care.
- The impact of IP policy, and the role of public-private collaboration, needs to be clarified for each of these three phases. For example, making the study data and tissue resources “open-access” and limiting private control over discoveries may promote secondary research use (objective 2). However, these same policies may hinder private incentives to invest in the research resource itself (objective 1) and may have a mixed effect on the number of discoveries that actually are translated into clinical use (objective 3). All of these impacts need to be considered. The Draft Report’s discussion of patent thickets (on p. 31, lines 1243-45) may merely have been intended as one example but, without more, it provides only a partial and one-sided view of these issues. IEEE-USA strongly believes these matters must receive fuller analysis before policy is set.
- Open-access to data does not necessarily imply that there should be public control of discoveries made using those data. The data and tissue resources created by this study would be very important inputs to research and discovery, but they are not the sole inputs. The proposed national database will produce clinical benefits only if additional inputs—*e.g.*, funding, insight, equipment, manufacturing capability—are expended on research and product development. Parties that expend those additional resources must be able to reap some private benefit from the resulting discoveries, while still recognizing the public’s role in creating the national database on which the research relied. Unless an appropriate balance is struck, the database may fail to mobilize needed private investment and may fail to deliver its hoped-for public benefits.
- The IP policy for this project needs to find a middle ground that appropriately shares the value of discoveries between the public and the private discoverer/developers. There are various well-known approaches (such as compulsory licensing or the use of collective rights organizations) that would allow private patenting but make it subject to public duties. Additional approaches might be developed by thinking of the national database as a public resource and drawing on traditional concepts, such as those seen in electric utility regulation, that require non-discriminatory access for all users, but charge different access fees to different classes of users. For example, academic/governmental users who are willing to place their discoveries in the public domain might be granted access to use the database “at cost,” whereas commercial users that wish to patent their discoveries could be granted access on terms that require them to pay higher access fees, pay a royalty on discoveries, or agree to grant limited access to their discoveries for non-commercial researchers. The national database presents novel issues and all options—including new ones—should be carefully explored. Appropriate statutory authority should be sought from Congress to

implement a framework that fairly apportions the value of discoveries made using the national database between the public and private interests, creates incentives for commercial translational research, but preserves needed access by non-commercial researchers.

Integration of Federal Agency Efforts

We believe that not only NIH but all the Agencies within the Department of Health and Human Services (i.e., AHRQ, CDC/ATSDR, CMS, FDA, HRSA, Indian Services, NIH, etc.) need to integrate their research-knowledge into a body of work and that this integration should include other Departments as well whose expertise relates to the public health. Examples of other agencies that should be involved include: the Environmental Protection Agency (environment / air and water), the US Department of Agriculture (food), the Department of Energy (nuclear and radiation), the National Oceanic and Atmospheric Administration (weather, global warming), the Department of State (global health), and the Department of Veterans Affairs (expertise in EHRs).

Previous IEEE-USA Work on Medical Technology Policy:

The IEEE-USA Medical Technology Policy Committee has worked diligently through the years in addressing many of the objectives that we support through the creation of Position Statements. The following position statements have been adopted by IEEE-USA's Medical Technology Policy Committee (these position statements can be accessed at <http://www.ieeeusa.org/policy/positions/index.html>):

- Addressing the Health Care Needs of our Aging Population With Technology (Nov. 2005)
- Interoperability of the National Health Information Network (Nov. 2005)
- Homeland Security Operations and Use of Personally Identifiable Health Information (June 2005),
- National Health Information Network, with Emphasis on Security and Privacy Issues (June 2005)
- Support for Bioinformatics Infrastructure and Technology (Nov. 2005), Home Healthcare Technologies (November 2004)
- Improving the Health Care System Through Use of Information Technologies (June 2002),
- Nondiscrimination in Employment Based on Genetic and Other Health Information (Aug. 2002)
- Privacy (Sept. 2003)
- Quality of Health Information on the Internet (Nov. 1999)
- US. Government Advisory Entities Concerned with Health Related Issues (Feb. 2002)
- Universal Access (Nov. 2003) and
- Voluntary Healthcare Identifier (June 2004).

On January 30, 2006 we, as a member of the Coalition for Healthcare Funding, sent a letter to President Bush and Members of Congress supporting increased federal funding for [public health research](#) [IT'S TIME TO MAKE PUBLIC HEALTH FUNDING A NATIONAL PRIORITY]. Later on March 14, 2006 another Coalition letter was sent through the Genetic Alliance to Dr. Mark McClellan, Commissioner, U.S. Food and Drug Administration to issue [regulations for a genetic testing specialty](#) under the Clinical Laboratory Improvement Amendments (CLIA) of 1988.

Within our Medical Technology Policy Committee, we have a Genetics and Bioinformatics Working Group which helped draft the June 18, 2003 letter sent to the Senate Committee on Health, Education, Labor and Pensions recommending additions to the [Genetic Information Nondiscrimination Act of 2003 \(S. 1053\)](#), and later the 21 April 2005 one, sent to the House and Senate sponsors indicating support for the [Genetic Information Nondiscrimination Act of 2005 \(S.306/H.R. 1227\)](#).

Because of the potential harmful use of genetically engineered and other pathogens we also sent a letter to Rep. Jim Turner (R-TX) expressing support for the [Rapid Pathogen Identification to the Delivery of Cures Act \(H.R. 4258\)](#). 22 June 2004.