

**Summary of Public Responses to a Request for Information on
Improving Health and Accelerating Personalized Health Care through Health
Information Technology and Genomic Information in Population- and Community-
based Health Care Delivery Systems**

May 2007

Introduction

In November 2006, the Department of Health and Human Services (HHS) requested input from the public on plans for developing and using resources involving health information technology (IT) and genetic and molecular medicine, with specific reference to incorporating these capacities in evidence-based clinical practice, health outcomes evaluations, health care delivery, and research. This document summarizes the ideas and concepts received from the public in response to that RFI. The purpose is not to provide the views of the Department, but rather to summarize the issues commenters raised in response to the RFI. The Request for Information (RFI), the submitted responses, and accompanying information are posted at <http://aspe.hhs.gov/PHC/RFI/index.cfm>.

Background

Advances in medicine, biomedical science, and technology development present opportunities for increasingly patient-specific health care practices, by taking into account individual differences in health states, disease processes, and outcomes from interventions. The objective of such “personalized health care” is to improve the effectiveness and safety of medical treatment and prevention.

The application of interoperable health information technologies (HIT) in the health care setting, particularly the electronic health record (EHR), provides new opportunities to collect and analyze information about diagnostic and therapeutic interventions, as well as health care outcomes. Integrated data analysis of multiple parameters of health care practices has the potential to support new approaches to evaluating health outcomes, developing the evidence base for best practices, identifying individual differences in response to therapies, supporting research on new interventions, automating the process of detecting and reporting notifiable disease conditions and health care-associated infections to public health surveillance systems, and enhancing patient safety.

Occurring in parallel with the advances in health IT is accelerating the application of molecular and genetic medicine. While still in the early stages of entry into health care, these advances promise to provide useful information to aid in patient care through more accurate diagnosis and treatment at an individual level. As family history-taking becomes a more rigorous part of health care practice, and as more patients undergo genetic testing, the inclusion of genetic information in the health record can provide opportunities for further evidence development, knowledge management and advancement of personalized health care.

Realizing these opportunities will require new capacity to aggregate and correlate data from clinical records with outcomes or other relevant findings from multiple sources. Many public and private organizations are engaged in planning for future collection and integration of health data, incorporating advances in genomic science. The RFI sought input to help understand current directions being taken and the productive role that Federal health agencies might play in facilitating progress, avoiding unnecessary barriers, overcoming policy and logistical challenges, and achieving optimal benefit from the opportunities afforded in achieving personalized health care.

Responses

A total of 61 responses were received from a wide variety of stakeholders including: individuals; policy analysis organizations; advocacy groups; diagnostics and pharmaceutical companies; consulting companies; academic medical centers; state, regional, and local governmental agencies; developers of health IT products; hospitals; health care payers; and professional societies. A number of issues of interest were identified in the RFI.

An overview summary of responses follows:

Ethical, Legal and Societal Implications (ELSI)

The issues of privacy and security of personal health information, as well as protection against genetic discrimination, were discussed by many respondents. Successful resolution of these issues was commonly seen as crucial to achieving the goals of personalized health care. However, while some comments focused on perceived gaps in existing or prospective protections, others emphasized that provisions intended to protect individual privacy might result in impeding aggregation of data and hence potential improvements in population health.

Comments focusing on the need for individual protections included:

- *Support for enactment of federal law to prohibit discrimination on the basis of genetic information in employment and health insurance.*
- *Comments on the adequacy of privacy protections provided under the Health Insurance Portability and Accountability Act of 1996 (HIPAA).*
- *Discussion of the ways in which personal genetic information might differ from other typical health information; and the assertion that these factors might justify greater-than-normal protection of personal genetic information.*
- *Concern regarding collection of population-based genetic information and the sale of such data, or other uses apart from health care.*
- *Discussion of the need for consumer/patient “buy-in” and consumer understanding of gene-based diagnostic findings.*

Comments addressing the balance between protection of personal information and the potential for public health improvement through health IT-supported data aggregation and other research included:

- *Suggested approaches for achieving public health and research advances while adequately providing protection of personal health information.*
- *Suggested revision and improvement of current HIPAA-mandated standards for privacy of individually identifiable health information.*

Other responses discussed potential new patient rights and control over their personal information as the key elements of personalized health care, especially health IT and personal genetic information, become routine in health care.

Education and Communication

A number of respondents highlighted the role of education and communication for both the health professions and for the general public, identifying these as key elements for bringing about adoption of personalized health care, especially genetic and genomic technologies.

- *Respondents cited the need for continuing education of health professionals regarding the application of medical genetic tests and information.*
- *Several pointed to anticipated delays in implementing genomic-based health care practices due to a shortage of physicians, pharmacists, nurses, and other health professionals with training in genetics and/or the expertise required to incorporate genomic information in their practices.*
- *Commenters also pointed to the special requirements and need for genetic counseling as genetic testing is increasingly adopted.*

Respondents also pointed beyond educational efforts to the creation of ongoing systems for providing information to clinicians about best practices and standards of care.

- *Comments cited a need for expanded efforts to translate scientific findings into clinical practice.*
- *Special attention was drawn to the potential for clinical decision support through health IT systems.*
- *Respondents noted that both the technological and analytical foundations for clinical decision support remain to be built.*
- *Comments also called for examination of liability and other legal issues that might arise from clinical decision support systems.*

Respondents also cited the need for efforts to enhance public understanding of genomic information.

- *It was recommended that knowledge management systems should be user-friendly so that health care professionals, policy-makers, and the general public alike can*

be aware of published scientific information for evaluation and use to change health care patterns, when appropriate.

Health Information Technology

Many respondents stated that the use of electronic health records as a tool for enabling portability, storage, and interoperability of personal health information will serve to make the current health care sector more efficient and effective. A number of health IT infrastructure, policy, and architecture issues were discussed. Most responders stated that in order to achieve personalized health care, linkages between internal and external databases will be necessary and that common standards for privacy and security must be in place.

A number of respondents referred to potential gains from implementing an electronic system.

- *They suggested that health IT will allow health care providers to take advantage of enabling technologies and diagnostics and facilitate the use of new discoveries for the personalization of medicine.*
- *Some respondents commented that health IT can help both patient and provider in meeting personal goals, meeting previously agreed on plans, and integrating nationally accepted medical recommendations and criteria.*
- *Commenters also said that health IT can enable access to data and serve as sources of clinically rich, patient-centric information for community and population data aggregation needs, as well as for recognition of correlations and trends.*

Respondents identified a number of barriers to the implementation of interoperable systems that would allow electronic exchange of health information. These included:

- *The need for standardization of laboratory and clinical terminologies.*
- *Inability of health information systems to integrate genetic molecular test management and reporting.*

Several respondents discussed potential approaches for the collection of patient data and considered the pros and cons of these approaches. In general, most respondents thought that health IT should function to facilitate research and discovery, efficiently connect patients with their health care, and provide a resource for clinical decision support to assist physicians in the application of personalized health care. Other considerations that respondents noted might help to achieve the goals of HIT include:

- *The need to support information management tools for protocols, inventory, sample processing, and access privileges, patient authorization, and anonymization of data.*
- *The need to develop integrated laboratory information management systems to support clinical and research environments.*

- *A need to enable electronic health records (EHR) to serve as a resource for collecting and disseminating standards and best practices, and to facilitate access to data and disease modeling.*
- *Turning to early adopters of EHRs as resources that serve as a starting point for collecting and disseminating standards and best practices.*

A number of respondents wrote that these goals will be difficult to achieve in the absence of a central governance body that can provide guidance and a clear, detailed set of standards and instructions for the designers and users of EHRs, as well as for the developers and users of searching and interconnecting tools.

Additional responses addressed the need for systems development across the health care enterprise. There were a significant number of responses regarding centralized and decentralized approaches to data management. Some of the points respondents made about these approaches included:

- *An integrated laboratory information management system (LIMS) is essential to support genomic IT.*
- *Remote data collection has, in several environments, moved past the research phase and been proven effective for clinical care decision making. In several discrete but substantial markets, the EHR model has been expanded to encompass the range of data from clinical collection to subjective patient self-evaluations. This broader collection will provide valuable insight not only into interventions for health, but into opportunities to educate patients on their own decisions that affect their health.*
- *Unlike approaches that require building and maintaining interfaces between and among all of the applications in an enterprise, the centralized approach integrates disparate systems at the normalized data model level. Further, because integrations take place at the data model level, application changes do not typically require complex and expensive re-architecture of the system.*
- *In contrast to the centralized approach, the decentralized approach maintains multiple, duplicate physical versions of the data in an enterprise. Many legacy EHR systems employ a distributed or 'hub and spoke' topology. According to commenters, this approach takes significant time and cost to implement and maintain, is inflexible in supporting system and user change, and – inherently – leads to limitations in analytical capabilities and complicates data quality issues.*

Standards

Many respondents commented on the need for standards development and harmonization to enable the broad goal of resource sharing. They suggested that standards and definitions should be vetted and agreed upon by a wide variety of stakeholders to ensure that multiple systems across the health care sector are interoperable and can exchange health information, and that:

- *Standards for EHRs should support the collection of data required for supporting clinical trials.*
- *Scalable data standards for genomic information, biobanks, and data repositories are needed for sharing, collaboration and synthesis of information.*
- *A means for integration with existing systems is needed.*
- *Gaps in vocabularies and standards should be identified and addressed to ensure interoperability between EHR systems and clinical data systems.*
- *Attention should be applied to harmonization with international standards.*

Respondents suggested that establishing standards for interoperable clinical data repositories and datasets of appropriately protected patient health information would provide a foundation for useful linkage of different databases of information. The result, they said, could be a richer more complete knowledge repository that could be used for effectiveness, safety, or quality research.

Several responses offered additional perspectives on the utility of interoperable databases. By establishing standards to develop interoperable clinical data repositories and datasets of appropriately protected patient health information, the groundwork could be laid for the linking of different databases of information. This could result in a richer more complete knowledge repository that could be used for effectiveness, safety, or quality research.

Many respondents identified the value of data integration and how integrating data from various sources will facilitate research discovery. However, in order to do this, they noted that data must be extracted from different environments, often with different formats. Several respondents had general concerns about linking databases of patient information, including:

- *Issues regarding the creation, accessibility and analysis of data from multiple databases holding genomic information.*
- *That there is a lack of standard formats for obtaining patient consent and institutional review board (IRB) approval at the outset of building a dataset*
- *Unresolved challenges still exist related to data ownership stored in centralized databases/repositories.*

The Patient and Genetic-based Diagnostic Tests

Many respondents discussed issues surrounding integration of genetics based diagnostic tests into routine clinical care. Specific responses made about this topic included observations that:

- *Genomic data should be integrated into medical decision-making when value can be demonstrated.*
- *Guidelines related to clinically acceptable use of pharmacogenetics information in patient care should be developed.*

- *Predictive genetic testing should be available to patients who request it based on a family as well as clinical history that warrants risk assessment for early prevention or disease management.*
- *For rare diseases and conditions, it is important to be able to aggregate data about as many patients as possible to obtain sufficient statistical power.*

Several respondents also expressed concerns about incentives for developing new diagnostic tests based on genetics, pointing to reimbursement policies that are not designed to reflect the value of these tests when linked to a treatment decision.

Respondents made specific suggestions concerning the effective collection and use of genetic information in the EHR. These included:

- *Molecular diagnostic tests for disease may be expanded to cover additional genes, as new genes and multiple gene interactions are implicated in disease. The exact regions of DNA that were sequenced in any given molecular test should be recorded, for future clinical decision support.*
- *Genetic determinations may benefit medical decision making not only for the immediate patient but also for the patient's relatives who may be at risk as well.*
- *Genetic testing and data collection should be combined with access to counseling, prevention and treatment, with full recognition that there may be attendant costs.*
- *For many common diseases (e.g., cancer, heart disease and stroke, and diabetes), risk prediction will depend on algorithms that include genotypes, environmental exposures and lifestyle factors. Family history is a currently available tool that reflects genetic susceptibilities, environment, and behaviors that can be used today to estimate risk and personalize prevention strategies.*

Databases and Repositories

Many responses examined the potential for linked databases containing patient information, biospecimens, and genetic profiles as a source of information for public health, as well as outcomes and effectiveness research.

Several respondents pointed to the role that longitudinal data collection could play in understanding the genetic and environmental factors that lead to disease.

- *Respondents highlighted factors involved when collecting longitudinal data, such as the consent process and standards for collection, processing, reporting, and storage.*
- *Comments also noted the further clinical research required for genotype-based phenotypic risk assessment in a broad spectrum of individuals.*
- *One respondent suggested that health IT-enabled longitudinal data repositories might bring a new kind of transparency into the health care realm, enabling manufacturers to monitor and assess the types of patients receiving their*

products, the methods of prescribing and utilizing products, and their associated outcomes.

Better integration of clinical trials information systems with routine clinical care, and use of prospective data for clinical research, were also featured in several responses. These respondents suggested that this information will:

- *Promote the development of personalized medicine.*
- *Promote safe management of patients as they undergo treatment.*
- *Reduce the costs of cohort selection, targeted sample acquisition, and analysis.*
- *Provide the ability to answer questions important to study and protocol design in real-time and virtually simulate a protocol's recruitment potential.*
- *Allow clinically-validated patient-genomic information to be added upon sampling/testing.*
- *Allow for tracking and managing clinical management and disease response, which are important for evidence-based medicine.*

Several responders suggested that the use of interoperable systems of near real-time, secured patient information would be a rich source of information for outcomes and effectiveness evaluation. For example, they said that these systems could support:

- *Compilation of clinical laboratory data (including anatomic pathology) for a defined patient population which provides high-quality data for analysis of frequency of patient care encounters, utilization of the medical system, and quality of care, and*
- *Capture of health care data from the clinical laboratory that supports effective management of patient populations for disease susceptibility, clinical course and outcomes, treatment response, evidence-based clinical decision support, and optimization of health care.*

The use of clinical informatics systems for public health research was also discussed by a number of respondents. They suggested that population studies are needed to:

- *Identify associations between genetic variants and disease, assess risk factor interactions (gene-environment and gene-gene interactions), and establish risk factor prevalence estimates.*
- *Identify disease trends at the population level, provide a mechanism for bioterrorism surveillance, help to measure quality, and facilitate research.*
- *Allow public health organizations to customize their efforts to target specific diseases more prevalent from one population to another.*

Respondents also provided discussion regarding the accumulation of biospecimens, including consideration of a possible central repository. Most respondents thought that biorepositories should be integrated and linked in electronic clinical systems, and further that:

- *Appropriate processes and infrastructure to handle biospecimens in an accurate and secure manner are needed.*
- *Biobanks can harness the power of both genomic and clinical data and serve as a critical bridge between basic and applied research, linking laboratory to patient and getting cures faster.*
- *Patient advocacy groups should be brought into the process as partners in helping to ensure that the patient community understands how biobanks work, and the role they play in the clinical research infrastructure.*

Policy-related suggestions were made in some responses to address the use of patient information for research purposes. In their comments, respondents indicated that:

- *HHS should harmonize the regulatory and policy requirements of its various offices and agencies, particularly the Office for Human Research Protections and Food and Drug Administration, and also the National Institutes of Health; among the harmonization issues that need to be addressed is the reporting of unanticipated problems and adverse events in clinical research.*
- *Progress in research relating to personalized health care and other areas of public health require better, more robust partnerships to ensure access to diverse populations that are truly representative and offer the broadest basis for observation.*
- *Federal legislation and public education on privacy and confidentiality are needed to provide individuals with protections from risk associated with genomic test information.*
- *Consensus should be developed among health care providers, the pharmaceutical industry, institutional review boards, and government agencies on the mechanism whereby de-identified samples can be collected, stored, and analyzed.*
- *HHS should revise and/or improve upon the current HIPAA Privacy Rule to address concerns regarding access to patient health information for legitimate health and medical research, and should further harmonize the HIPAA Privacy Rule with the human subjects protections provided under the Common Rule.*

Effectiveness and Economic Analysis

Respondents suggested that adoption of new HIT and genomics based diagnostics in the health care system should be supported by evidence that improvements in the safety and effectiveness will result. Several commenters discussed the resources and models of comparative effectiveness and economic analyses required to support widespread adoption. Their comments included the following:

- *The costs of implementing interoperable HIT systems are high for small to mid-size hospitals, independent physicians, and smaller group practices.*
- *Incentives should be aligned to facilitate data sharing and institutions must be motivated to do so.*

- *A quasi-government body or collaborative organization on a national level could set priorities and direct or fund test and drug development. It could conduct comparative clinical and cost-effectiveness research on new and existing procedures, therapies, drugs, medical devices and other technologies, but it likely would not have any role in setting coverage or reimbursement policies.*
- *If therapies could be directed much more efficiently to those who will actually benefit, the resulting savings to health care system would be enormous.*
- *Centers for Medicare and Medicaid Services (CMS) should implement increased reimbursement rates for health care providers that adopt EHRs at the point of care and decreased reimbursement rates for health care providers that do not adopt EHRs at the point of care.*
- *The implementation of a drug safety clinical genomic system may provide the best overall return on investment for the health care community in the near-term. This is because the system would utilize single nucleotide polymorphisms (SNPs) that have an established link to drug safety outcomes and therefore can include information-based guidance to patients that possess SNPs relevant to drug safety, exploit a prescription/dispensing system already guided by an information system, and provide a cultural shift in pharmaceutical drug development whereby new drug indications can require genomic screening to increase the overall safety and efficacy of new drug entities.*
- *Medical advertisers are willing to support diagnosis search tools, as they have supported professional journals through print advertising. Using such approaches, diagnostic software may be kept free to users, using some combination of search-dependent sponsored links and more general forms of sponsorship or grants.*

Acknowledgment

HHS appreciates the time taken by respondents to share their experiences and thoughts in reply to the RFI. The information and perspectives submitted will be useful to the Department as work continues to bring about the elements of personalized health care.

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