HIM and the Path to Personalized Medicine: Opportunities in the Post-Genomic Era

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HIM professionals have a lot to offer genomics research. Those interested in developing their skills in the sciences can dig deeper into this fast-emerging field.

Successful mapping OF the human genome, completed in 2003, ushered science into what is called the "post-genomic" period. In this era, genomic profiling is making advances in personalized health more rapidly than what one would have once thought possible.

Genetic information does not signal the advent of personalized medicine, but it is a new tool expected to aid in the understanding of individual disease risk and the development of strategies for individual health management. The impact of genomic information on the HIM field is tremendous and will continue to grow. For HIM professionals interested in helping advance the use of genomic information, many of the needs reflect traditional HIM skills.

HIM professionals have opportunities to contribute to personalized medicine their knowledge of healthcare delivery; health data law, management, statistics, and information technology; and coding and classification systems.

HIM's Past Role in Personalized Medicine

HIM professionals have always been on the path to personalized medicine. When a patient is admitted to a hospital, a patient-specific record is created and stored. If the patient returns for continued care, the previous record is retrieved for reference in providing ongoing care. Documentation of a patient's visit to a facility is added to the record, creating a longitudinal record of the patient's personal health history.

Thus HIM professionals have been the curators of one of the largest existing databases ever: the patient record. They have kept access to the patient record secure, maintained the privacy of its contents, and coded and abstracted patient information for the purposes of healthcare statistics and research. As the health record evolves from paper to electronic form, HIM professionals continue as its guardian and are growing as a profession to meet the needs and demands of this new technology.

The move from paper-based health records to electronic health records (EHRs) is a major step in improving personalized care. EHR development is facilitating the materialization of the health information exchange (HIE) that will, in turn, allow the development of a more complete patient medical history by linking patient information from various healthcare providers.[1] Use of the personal health record, maintained by the individual, will also improve individual care as patients may maintain medical information about themselves, apart from a provider medical record, that can greatly improve their own healthcare.[2]

Collaborative Opportunities with Researchers

The **Human Variome Project** is one genomics project that would benefit from HIM contributions. The HVP will eventually attempt to link its databases of clinically relevant mutations and genetic variations to patient health records and would therefore benefit from knowledge of health information technology procedures.

The project is an international collaborative effort to classify human genetic variation as it associates with phenotypic variability and human disease indicators. A centralized database, with centralized governance, will link knowledge obtained from clinical, medical, and research laboratories.

A co-requisite of developing a database such as this is the development of data standards and a standardized nomenclature of human genetic variation to enable data sharing and interoperability.[3] Since the HVP seeks the ability to exchange genetic information related to health, components of its database can contribute to the development of a national HIE.

To facilitate the merging of patient phenotype data in an HIE with information in genetic databases, HIM professionals can collaborate with scientists on database construction. Although many healthcare system databases are still in the process of transitioning from paper to EHRs, the patient healthcare record database remains intact. Patient data continue to be added to the database, but the fundamental structure of the database remains unchanged. The focus on health record databases is concerned not with how to construct a database, but rather with the issues associated with converting to a different type of media upon which to store health information in order to facilitate the linkage of provider databases.

Academic researchers are still constructing their databases and facing challenges such as developing methods for entry validation and authentication of research data. HIM professionals possess knowledge of clinical documentation practices such as authentication, amendments, and audit trails that can be of aid to research scientists who are developing databases to link genetic variation with disease phenotypes.

Research groups are working on methods to classify human genetic variation based on disease.[4] These efforts, as a consequence of databases successfully linking genetic variation to disease phenotypes, could potentially lead to a disease coding classification system such as a future version of ICD-CM that will have an axis to incorporate human genetic variability data.

HIM procedures for de-identification of data may also be of interest to the scientific community—for example, how data are abstracted for the compilation of disease, physician, and operation indices that are later used for in-house quality assurance and epidemiological studies; how billing data are abstracted for financial reimbursement and utilization review; and how birth certificate data are abstracted and forwarded to the National Center for Health Statistics to study birth events and pregnancy. Abstracted, de-identified genomic data will be essential for research studies that seek to identify risk factors that may affect personalized health without compromising patient privacy.

Another database that may link to health records, and would therefore also benefit from HIM knowledge and skills, is the **nutritional phenotype database** (dbNP) being developed by an international network of scientists under the auspices of NUGO, the Nutrigenomics Organization.

This database is being designed to capture, store, and manage biological information and primary data from the results of nutritional research. Not unlike the HVP, it too will need to develop standards for nutritional data in order to enhance interoperability among the data sources that feed into the dbNP.[5]

Databases such as these are being constructed to link to genomic databases and address particular health issues. One of the goals of the dbNP project is to link the results of small independent research studies, allowing for knowledge building and improved data consolidation and interpretation.[6] These efforts are likely to be translated into improved personal and public health.

For example, NUGO and its international partners hope to address issues such as obesity. Scientists in nutritional research are focusing efforts to develop new tools and accurate methods for analyzing an individual's nutritional needs and monitoring an individual's nutritional intake.

Although an individual's amount of nutrient intake is critical for maintaining one's health, a re-emerging awareness is that individuals differ in metabolism. Therefore, an individual's genomic makeup must also be analyzed, because an individual's genes determine the activity of proteins that function in cellular metabolism.

The interaction of nutrients with an individual's genetic makeup plays a significant role in affecting individual risk for disease. These gene-environment interactions underlie health and predispose one to maintain health or develop disease. Phenotype data from health record clinical lab reports, as well as nutrient intake data, will provide additional information for improving personalized health for obese individuals.

Database Challenges

A sense of how difficult it is to develop a database such as the two mentioned above can be captured in the following statement made by Warren Kibbe, cofounder of the NUgene Project, a genetic banking study sponsored by Northwestern University's Center for Genetic Medicine in partnership with four other institutions:

> "Pharmacogenomics and nutrigenomics biobanks in particular present some difficult issues such as properly gathering and annotating disease and nutrition information for each participant, designing consents and procedures (and obtaining proper approvals) for discovery banks (versus a hypothesis driven bank), and navigating the ethical, regulatory, funding, and intellectual property ramifications of a genetically focused bank. Add to the mix the complexities of handling and tracking, and finally providing robust phenotype mining tools and analytics for genotype/phenotype association studies and you get a sense of the potential complexities underlying the establishment of a nutrigenomics biobank."[7]

Several ways HIM professionals could help contribute to the development of a biobank are in designing, creating, and implementing:

- Consent forms that allow for continual mining of patient information from EHRs, billing records, and ancillary documents
- Data collection forms from self-reported information such as patient information from PHRs so that the information collected meets standards for data sharing and interoperability
- Policies and procedures to protect the identity of patient forms and biological samples and to track their workflow trail
- Policies and procedures for de-identifying patient data (from biological samples, clinical measurements, billing information, and dietary and epidemiological data) and recombining de-identified data for use later in statistical analyses
- Policies and procedures that ensure authentication of data entries in the system, the existence of appropriate functions for amending and auditing data, and the establishment of record retention guidelines
- Policies and procedures for tracking and auditing data that are obtained from ancillary or follow-up studies and linking it to previously collected patient data
- Policies and procedures to authenticate and audit access to biobank data and to ensure that access is given only to authorized individuals in concordance with the needs and responsibilities of their position as well as revoking privileges of those same individuals should they abuse their right to access
- Policies and procedures to train individuals to use the biobank and to properly use the system[8]

The state laws and federal regulations that govern the operations of HIM departments will also have a role in guiding the establishment of regulatory principles governing biobanks. Patient data housed in a biobank's computerized systems will be subject to the same privacy and security measures to which EHRs are subject. HIM knowledge of HIPAA and the development of the Nationwide Health Information Network can also provide additional guidance for the establishment of a regulatory framework for biobanks.[9]

In designing a biobank's system architecture, the desired goal is the clear demarcation of the data model, the business/scientific model, and the user interface. The system architecture must ensure that the biobank is highly secure yet flexible, is regulation compliant, has programmatic interoperability, and that common data and exchange standards are used. Patient health identifiers must be kept entirely separate from de-identified patient health and billing data and databases connecting to the biobank also must maintain various levels of de-identified data.[10] HIM professionals can use their experience with EHR databases to assist in all of these aspects of biobank database management.

Patient Privacy and the PHR

There are many issues regarding patient privacy in the post-genomic era.[11] Related is the issue of PHRs, which will test HIM skills in protecting patient privacy.

As technology improves and the cost of genetic testing gets cheaper, more and more individuals may decide to get tested and incorporate their genetic test results into their PHR. Providers have a professional responsibility to review the information contained in a PHR and determine if any of it can be used to improve the patient's care.

Policies and procedures must be in place to prevent the risk of inadvertent disclosure; without them, patients may withhold vital information from the provider.[12]

As with biobanks, HIM professionals can assist in developing policies and procedures for obtaining self-provided information and protecting its privacy.

Statistics

Many opportunities are appearing for those individuals who enjoy data analysis and statistics. Bioinformatics, genetic epidemiology, statistical genetics, and statistical methods development will be in demand, and analyzing complex genetic data will require individuals with deeper statistical knowledge.

Current statistical models describe population behavior. The goal of personalized medicine, however, is to combine genomic, proteomic, and metabolomic data of individuals to better characterize the population into more homogenous subgroups with respect to factors that influence individual disease risk. Doing so will enable statistical models and methods that can more accurately predict individual disease risk. One example that illustrates the use of genomics in advancing personalized medicine is in population admixture mapping.

Population admixture occurs when a population represents two or more subpopulations that have differential allele frequencies for a gene.[13] The idea of admixture mapping is not new. But in the past, statisticians have not been able to fully harness its power for discovering genes that contribute to the expression of complex traits.

Because of the explosion of genomic information, however, statisticians are now better able to develop statistical tests that can be used to analyze data from admixture-mapping studies to discern allele frequencies of subpopulations that may indicate that a particular subpopulation is at risk for disease.[14,15]

Analysis of high-dimensional data (such as that applied to analyze gene expression data obtained using microarray chip technology) is not just limited in application to genomics studies. Other facets of personalized medicine such as financial management also have large, complex data sets of interest to an individual's health.

One such example is the application of dimensionality reduction to data mining methods and Bayesian classification models in order to identify risk factors for health plan patients who enroll in a disease management program for their substance use disorder (SUD). Models were developed to target patients to an appropriate level of SUD management based on individual risk of SUD relapse and simultaneously optimize the utilization of healthcare resources.[16]

HIM professionals not interested in genomic studies can still apply the principles used to analyze high-dimensional genomic data to the analysis of other types of data.

To become more effective in data analysis, HIM professionals will need to acquire or develop advanced statistical skills. This will require a strong background in the mathematical sciences up to and including calculus, linear algebra, and probability theory.

Health IT has been and remains an interdisciplinary field. In the past, health record professionals have primarily interacted with individuals practicing medicine. That circle now is expanding to include scientists, statisticians, and information technologists. Even HIM professionals in roles other than data analysis may still require enough statistical knowledge to work with data analysts or statisticians who will access health record data for genomic studies.

Possible settings and opportunities to network for individuals who choose to get involved with personalized medicine include academic and governmental research institutions that engage in personalized medicine research. Some of the new skills HIM professionals need to develop and current applicable HIM skills in the post-genomic era are provided in the sidebar below.

Skills for HIM Professionals in the Post-Genomic Era

Current Skills to Apply

- Policy and procedure development for managing patient health information to ensure its accuracy, integrity, privacy, and security (including investigating and resolving problems that may arise in the development of phenotype and genotype databases that involve breaches of patient information).

- Knowledge of EHR database systems design and their maintenance.
- Knowledge of forms design, computer input screens, and clinical documentation tools and guidelines.
- Knowledge of implementing industry standards for data sharing and interoperability.
- Developing standardized healthcare data sets.
- Knowledge of coding and classification systems.
- Experience with organizational compliance to laws, rules, and regulations for licensure and accrediting agencies.
- Knowledge of maintaining a master patient index and master client index.
- Developing policies and procedures for release of medical information.

Skills to Develop

- Statistics for data analysis, including calculus-based probability and statistics; statistical programming languages (SAS, S-Plus, R)
- Information science
- Computer science
- Knowledge of genetics

The power of combining research and health information technology to advance personalized medicine will be witnessed in projects like eMERGE, the Electronic Medical Records and Genomic Network, a consortium of biorepositories that are linked to electronic patient health records for the purpose of conducting genomic studies.[17] Projects like these will serve as prototypes for developing databases now and in the future. HIM professionals collaborating with researchers will push the progress of personalized medicine faster to the goal of improving individual patient care.

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Notes

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