Genomics and HIM: Three Areas of Increasing Intersection

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The pace of genomic discovery since the 2003 completion of the Human Genome Project has been breathtaking. Today scientists are making rapid inroads on new approaches to prevent, screen for, and treat some of the most important conditions affecting the public.

Prior to 2005 these discoveries largely related to less common classical genetic conditions such as cystic fibrosis and sickle cell disease. Since that time there has been a burst of discoveries relating to common complex conditions—those in which genes and environment play a major role—including heart disease, diabetes, and cancer.

The cost of obtaining an individual's genetic blueprint has also dropped dramatically over the last five years, and within a decade it is likely that a patient's entire genetic code might be sequenced for around the cost of an MRI scan today. As a harbinger of this, commercial entities are currently offering "genome-wide scans" that measure hundreds of thousands of genetic variations associated with disease risk in individual customers for a few hundred dollars.

Often these tests are offered without the involvement of the consumer's healthcare provider. Not unexpectedly, such tests have been the subject of considerable debate in the scientific community.[1]

The unprecedented accessibility of genomic information has fueled a growing interest in personalized medicine. The central tenet of personalized medicine is that knowledge of an individual's genetic blueprint can improve the current "one-size-fits-all" model of healthcare delivery by using genetic information to better tailor health interventions.

Clinically accepted examples of personalized medicine can be found in the field of oncology, where genomic insights are being used to guide personalized therapeutic approaches for a wide variety of malignancies.[2]

Not surprisingly genomics and personalized medicine have become caught up in the ongoing healthcare reform debate. Though it is too early to determine if personalized medicine will be the savior or scourge of the US healthcare system, it is clear that genomic information will play an increasing role in healthcare delivery.

HIM systems will play a pivotal role in the successful integration and utilization of genomic information in healthcare. IT infrastructure, clinical decision support, and research are important to the intersection of genomics and HIM.

Infrastructure

It is fortunate that the push for robust and interoperable health IT systems coincides with the explosion of genomics in healthcare. Groups like the now-disbanded American Health Information Community's Personalized Health Care Workgroup have spent considerable time and effort laying the groundwork for integrating genomic information into health information systems.

It appears that the Office of the National Coordinator for Health Information Technology will continue to work to integrate genomic information into health IT systems. Ongoing attention to the development of genomics-enabled health IT will be crucial to realizing the full potential of genomic information in healthcare.

Genomic information poses a host of interesting challenges to information management. The first of these may simply be storage capacity. If sequence data for an entire genome become part of the individual's medical record for life, health IT systems will need to be developed that can store enormous amounts of data securely and offer access to that data across that person's life span.

It is likely that an individual's record may need to contain more than a single genome. For example, an individual with cancer may need to have both the tumor genome and his or her "normal" genome available to optimize therapy.

It also is critical that systems offer interoperability of data over a prolonged time period. Genomic information may be the ultimate example of legacy healthcare data—the primary sequence of an individual's genome at birth will follow the majority of his or her cells to the grave.

It is very likely that it will become economically viable to sequence an individual's entire genome once and then access relevant bits of information as needed during the lifetime of that individual. Under this model, primary sequence data should be portable across multiple health IT platforms.

Systems also must be capable of re-analyzing the primary sequence data when new information about how to interpret a given sequence becomes available. Thus it will not be enough to simply store static interpretations. The healthcare industry will be learning about the genome for many decades to come, and the meaning of any given genomic sequence may change over time.

Finally, systems need to be designed so that genomic information is afforded the same level of security as other sensitive parts of the health record. "Genetic exceptionalism" is a term used to describe the idea that genomic information is qualitatively different than other health information—and therefore requires protections above and beyond all other data in the medical record. In fact, confirmed abuses of genomic information are extremely rare.

The recently enacted Genetic Information Nondiscrimination Act (www.genome.gov/24519851) should further reduce the possibility of abuse.

Clinical Decision Support

If obtaining methods to inexpensively sequence the human genome was a challenge of the first decade of the twenty-first century, interpreting that sequence will be a challenge for the second decade and beyond. The busy clinician—no matter how expert—will be unable to keep track of the genomic information relevant to patient care.

An ongoing project at the National Human Genome Research Institute provides an early glimpse of just how complex this task may be. The project, known as ClinSeq (www.genome.gov/20519355), seeks to fully sequence a group of about 200 to 400 cardiovascular risk genes in individuals that have been risk-stratified for cardiovascular disease using conventional clinical information.

As of May 2009, more than 300 individuals had been sequenced for variations in approximately 200 different genes. Ten thousand distinct variations that potentially contribute to disease risk have been identified.

The ability to keep track of the health consequences of such diverse genetic variations for even a single class of disorders suggests informatics and effective genomic data management will play a crucial role supporting clinical decision making.

Health IT systems will need to be empowered to do much of the interpretation of genomic variation for healthcare providers. Clinicians will probably not need to know much about what is going on "under the hood" as long as a trusted source is producing and vetting the interpretive algorithms.

There is no widely accepted mechanism for developing, vetting, and disseminating such interpretive algorithms in the US today. Artificial neural networks constitute a developing set of decision-making tools that have been applied to genomics research since the 1990s. Their ability to observe large amounts of information and develop models that can interpret new data has great potential as an approach to managing genomic information in healthcare systems.

Research Use of Data

The following paradox faces those interested in applying personalized medicine in an era in which healthcare providers are exhorted to follow the precepts of evidence-based medicine: as one tailors treatments to an individual with multiple genetic variations and environmental exposures it becomes increasingly difficult to generate robust evidence of benefit.

This is a simple consequence of probability—the more factors that contribute to drug responsiveness, for example, the less likely it is that a large number of individuals will share all (or even most) of those factors. Developing sufficient evidence to support use of an intervention will require new methods for assembling and monitoring large numbers of individuals—and traditional ways of assembling research cohorts will likely be too expensive.

One potential solution would be to develop creative ways to harness IT, including data residing in EHR and PHR systems, to conduct certain types of research in real time. An example might be a study in which large numbers of individuals diagnosed with type 2 diabetes with known genotypes are monitored for how genotype affects responsiveness to various oral diabetes drugs chosen by individual healthcare providers.

There are emerging models where Web-based social networking approaches are being used to develop self-assembling groups of individuals interested in advancing research by volunteering the use of their health information online. To date these communities have relied largely on self-reported clinical information rather than information directly accessed from health IT systems.

Closing the loop between these communities and PHR and EHR systems could provide a powerful tool for advancing the understanding of genomics in healthcare. Pilot projects to explore the use of electronic medical records to support genomics research are under way in the Electronic Medical Records and Genomics Network (eMERGE), funded by the National Human Genome Research Institute and the National Institute of General Medical Sciences. Careful attention will need to be given to the ethical implications of such novel approaches to human subjects research.

Numerous issues will confront the growing intersection between genomics, personalized medicine, and health IT. Undoubtedly there will be vigorous debate on a number of these topics—and very likely all will be viewed through a lens of value offered to the US healthcare system. Those with extensive experience in HIM should be at the table for these debates. Fully informed solutions to these issues could yield tremendous benefits to individuals and society for decades to come.

Notes

1. Khoury, MJ, et al. “The Scientific Foundation for Personal Genomics: Recommendations from a National Institutes of Health-Centers for Disease Control and Prevention Multidisciplinary Workshop.” *Genetics in Medicine* 11, no. 8 (Aug. 2009): 59–67.
2. Chin, Lynda, and Joe W. Grey. “Translating Insights from the Cancer Genome into Clinical Practice.” *Nature* 457, no. 7187 (Apr. 3, 2008): 553–63.

References and Resources

National eHealth Collaborative. Available online at www.nationalehealth.org.

National Human Genome Research Institute. Available online at www.genome.gov.

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