

# An Early Warning

## The -omics Data and Personalized Medicine Are Coming

**D**URING THE COLD WAR, when the United States feared an attack by the Soviet Union, our radar constantly scanned the horizons for airborne objects headed our way. Any unexpected or unknown blip on the screen was cause for serious concern. Satellite surveillance ultimately provided us with an early warning system to prepare against a potential threat. In other words, we could see beyond the horizon and anticipate threats rather than waiting for them to materialize on our radar screens.

This column issues an early warning of a major change that is rocketing toward healthcare IT. The timing of its arrival cannot be predicted with certainty, but it is already visible in research reports published leading clinical journals. A few prominent medical centers are starting to incorporate this revolutionary change in selected clinical services, and private investors are already developing businesses that will be ready to commercialize it upon arrival.

The “missile” accelerating toward our horizon is a cluster of brand new medical services made possible by the Human Genome Project (HGP). It is analogous to an intercontinental missile with multiple warheads that will head in different directions before impact. No matter where these clinical bombshells land, in hospitals or outpatient sites, they will generate unprecedented volumes of new data. IT professionals need to start anticipating these changes that are just over the horizon and headed our way.

### GENOMICS AND PROTEOMICS AS EXPLOSIONS IN MEDICAL KNOWLEDGE

We already know that many diseases are rooted in genes. Cancer, heart disease, metabolic disorders, dementia, depression and many other serious health problems tend to run in families. Thanks to new technologies that quickly map the 6 billion base pairs (one massive data set!) of the four basic molecules that provide the blueprint for biological activity in each of our bodies, medical scientists can

now compare the genomes of healthy and unhealthy people. This testing is still too expensive for commercialization, but prices for genetic sequencing are falling fast.

Various combinations of genetic differences associated with disease—called single nucleotide polymorphisms, or SNPs (“snips”)—are now being used with stunning precision to differentiate life-threatening conditions from less-serious forms of several diseases. Medical science and bioinformatics are starting to identify therapeutically relevant variations in diseases at levels undetectable by visual examination.

For example, breast tumors that appear to be identical on a mammogram or a microscope slide can be very different at a molecular level. (Molecular expressions of genetic structure create the proteins that control cell function. This functional extension of genomics is called proteomics.) Armed with this knowledge, specialists can select the prescription drug or biological agent that targets the molecules of the patient’s specific tumor. This new ability to see disease at the level of the gene and protein will replace “one size fits all” medical care with personalized medicine.

Only a few dozen diseases can now be treated from this totally new and very promising perspective. Personalized

medicine as a universal standard of care is still over the horizon, not yet on the radar of forces that need to be addressed right away. However, it will quickly enter the realm of daily practice as the costs of accurate genetic and molecular diagnostics approach today’s prices for imaging and laboratory tests. Many experts expect the price of reliable genetic and molecular testing (e.g., laboratory on a chip) will fall to \$1,000 within the next two to four years. New types of data—and lots of it—will need to be collected, stored, and used when molecular medicine finally lands in the realm of everyday practice.

### NEW IMPORTANCE FOR OLD GENETICS

Ironically, the full-scale arrival of personalized medicine will likely draw unprecedented attention to an underappreciated realm of traditional medicine. Most large hospitals have a few genetic counselors and maybe a medical geneticist who labor in the background, consulting with special-

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ists whose patients may have an inherited disease (e.g., Huntington disease, reproductive systems cancers). These genetic specialists have worked for years with detailed family histories in text format and family pedigrees presented as diagrams.

Although personalized medicine in the future will provide direct evidence of diseases that have been suggested indirectly by family histories in the past, the study of disease development in a patient’s family will continue to be important. An individ-

ual's genetic heritage suggests general predisposition for many diseases. However, it does not mean he or she will necessarily get a disease that runs in the family. Ironically, the proliferating knowledge base of genomics is highlighting the importance of cultural, behavioral, and environmental factors that influence the development of genetic diseases.

The role of non-genetic factors in initiating and promoting genetic disease has been clearly demonstrated for many years by studies of identical twins. If one identical twin develops a gene-linked disease and the other does not, genes alone do not explain the problem. The new -omics data bases will need to include information about the cultural, behavioral, and environmental factors that define which individuals actually develop diseases to which they have genetic predispositions. Ironically, these historical data have been collected for years by genetics specialists, but they have not yet been incorporated into the electronic medical record.

#### **OBJECTS MAY BE CLOSER THAN THEY APPEAR**

Recent modifications of clinical data bases, such as adding procedural documentation to support pay-for-performance (P4P), will be relatively minor compared to the information-intensive scope of molecular medicine. Market forces have not yet forced enterprise EMR vendors to deal with the data of genomics and proteomics. Only automated systems will be able to handle the massive data sets of genomics and proteomics, so vendors face a challenge and opportunity.

Ironically, this coming revolution in medical informatics may initially favor growth of personal health records controlled by individual patients. We should be prepared for such surprises in its impact, but we should not be surprised when the genetics revolution hits health care. It's just over the horizon and headed our way like a rocket. **JHIM**

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# The Advent of RHIO 2.0

## The Country's Strategy of Creating Clinical Data Exchanges Is About to Undergo a Difficult Shift from RHIO 1.0 to RHIO 2.0

### **RHIO 1.0**

The creation of regional clinical data exchanges is a centerpiece of the U.S. national healthcare information technology strategy.

These exchanges are generally seen as having a regional master patient index, a service that identifies the various organizations that have clinical data about a patient and a mechanism to securely retrieve that data and present an aggregate clinical picture to a patient's provider. In addition to regional clinical data retrieval, these exchanges are also seen as supporting capabilities such as e-prescribing, bio-surveillance, quality data reporting and clinical messaging.

The Regional Health Information Organization (RHIO) is viewed as the organization that will oversee the creation and management of a clinical data exchange. RHIOs are a collaboration of regional healthcare stakeholders who come together to address issues of governance, funding, privacy policies, data sharing agreements and the management of the exchange technology infrastructure.

A National Health Information Network (NHIN) is envisioned as a technology, standards and management approach to integrating regional clinical data exchanges across the country.

One might call this vision RHIO 1.0.

Many communities have risen to the challenge of implementing this vision. While the exact number of RHIOs is uncertain, there are at least 200 such efforts across the country. The federal government has launched initiatives to establish interoperability standards, examine variations in state privacy laws, conduct

demonstrations of the NHIN and fund studies of areas such as strategies for state governments. Organizations such as the eHealth Initiative and the Markle Foundation have been exceptionally effective at bringing together the diversity of healthcare stakeholders and communities to share experiences, create tools and identify policies and steps that will facilitate the achievement of this vision of RHIO 1.0.

### **PROBLEMS WITH THE VISION**

The country has been pursuing this vision for almost three years. The experiences of many over this time suggest several significant problems with this vision.

Many communities are unable to come together in an effective, collaborative way. There are several reasons for this—intense regional competition, disinterest by key stakeholders and overly fragmented or large, local healthcare markets. These communities lack sufficient social capital or ability—the essential building blocks for the creation of a RHIO. These communities may have never worked together and as a result they have not forged the necessary effective working relationships or trust.

Even when that social capital exists, those who form a RHIO must overcome a series of challenging technical and non-technical challenges. Issues such as patient identification techniques, security, governance and privacy are formidable barriers and many communities are struggling to overcome them.

The creation of a clinical data exchange requires capital, at times significant capital, and means to support the financial requirements of ongoing operations.