



Vendors Ready Second-Generation Electronic Health Systems to Link Genomics and Clinical Records

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Several large healthcare IT providers are looking to get in on the ground floor of the translational medicine market by adding new capabilities to their systems to enable the integration of genomic information and clinical data.

GE Healthcare, for example, is developing a "clinical knowledge platform" called Qualibria that can "correlate genomic information with the clinical experience or the phenotype," Brandon Savage, chief medical officer of the firm's integrated IT solutions business, told *BioInform* this week. The system is scheduled for limited release to early-access users later this year and general release in 2011.

Cerner is also "trying to make it easier to connect" genomics and clinical medicine, said Mark Hoffman, vice president of life sciences solutions. One way the company is addressing this challenge is through the development of a Clinical Bioinformatics Ontology that links vocabularies used for genomics and clinical practice.

A number of academic medical centers are actively working at the intersection of genomics and the clinic, but these groups to date have not had much support from commercial systems.

For example, Lynn Vogel, vice president and CIO of MD Anderson Cancer Center, said in a keynote address at the Conference on Semantics in Healthcare and Life Sciences earlier this year that "commercial vendors in this space have had almost no interest in bridging the gap between the clinical side of the house and the research side of the house." As a result, Vogel noted, many translational research groups have had to build their own informatics systems to handle data from the two disciplines.

GE's Savage agreed that institutions like MD Anderson currently "tend to outpace the average vendor in the field" when it comes to fully integrated systems. "Their frustration is not surprising," he said.

"Historically, vendors have not had the right infrastructure to be flexible, but with this investment in [Quilibria], we do believe we will be significantly more responsive to these types of requests for research," he said.

Savage noted that electronic systems for research and the clinic are "typically very separate" in US healthcare institutions, but noted that the company is "investing a very large amount of money right now" in building systems that will address that challenge. He did not offer details on the investment sum.

Models and Ontologies

GE Healthcare has been developing Qualibria in collaboration with Utah's Intermountain Healthcare and the Mayo Clinic.

Savage said that GE is working with these academic partners to build "clinical data models" and to create a "public-domain" medical vocabulary for storing clinical information. So far, the partners have developed approximately 4,000 models that will help with data storage and "reliable" data retrieval for clinical research, he said.

He noted that GE's system is looking to improve upon existing medical ontologies such as SNOMED, the Systemized Nomenclature of Medicine Clinical Terms, and LOINC, or Logical Observation Identifiers Names and Codes, which are not organized "to store information about a patient in a way that is clinically relevant to a physician."

Having the models in place will make it easier to store and extract information and help "correlate genomic information with the clinical experience or the phenotype," Savage said. The company is also looking into statistical correlation mechanisms, for example, how SNPs representing disease states can be tied into a patient's electronic medical record.

Quilibria will include a "repository of all of the clinical information available on a patient, all the phenotypes, all of the disease experiences," including genomic information, Savage said.

It will be deployed with beta partners, including Intermountain and Mayo, later this year. The system will interface with GE's Centricity line of healthcare IT products and will also work with third-party electronic medical records, Savage said.

Cerner's Hoffman noted that while very few genetic associations described in the literature have found their way into clinical practice, that will change in the coming years. Once a physician decides there is clinical value in a test, genomic information will need to move around hospital IT systems, he said.

Users can already load SNP, copy number variation, and indel data into the current Cerner system, he said, but "don't always know they can." He admitted that the Cerner system is "not today" readily able to link complex clinical and genomic information, which is why he and his colleagues are developing new functionalities.

One hurdle, he said, is "siloeing within these organizations," and the fact that "there's not always good communication" between the research and clinical groups.

One project the company has underway to address this issue is the Clinical Bioinformatics Ontology, which has been curated at Cerner in line with suggestions from customers.

Hoffman chairs the ontology effort, which aims to close gaps in existing medical vocabularies and covers molecular genetics, molecular pathology, cytogenetics, and infectious disease.

The ontology currently has 14,000 concepts, which are "linked semantically," Hoffman said. For non-commercial purposes, Cerner grants users a royalty-free, perpetual right and license to use the ontology.

Applying this vocabulary, Cerner users can generate data without having to modify their electronic medical record schema, he said, since "they just tag a result with a concept identifier from that vocabulary," Hoffman explained.

Users can enter lab report data in a spreadsheet format or have it uploaded directly from an instrument. A spreadsheet row might include a lab test data point, another might be a clinically relevant mutation result, while the columns indicate the vocabulary terms, he said. "Behind the scenes, the results are tied to a concept," he said. For example a value might be tied to a LOINC code, another might link to an ontology concept, all of which are steps that make the data computable.

Over the last year, Cerner has focused its ontology efforts on cytogenetics, which Hoffman described as the "orphan step-child" in informatics. The company developed a method to parse karyotypes into "very discrete concepts," making them machine-readable. One next focus, he said, is on sequence annotation, for which "we want to improve our capabilities."

Hoffman believes that vendor systems offer a number of capabilities that are helpful for translational researchers, for example "the security we wrap around clinical data."

Security issues can be a big challenge in this space, he said, and in some cases have led clinical colleagues to exclude researchers from looking at their data. "We can do the scrubbing of the HIPAA identifiers" to facilitate communication between research and clinical domains, he said.

Do-It-Yourself

New offerings from GE and Cerner may address criticisms some clinical researchers have leveled against commercial electronic health systems, which many have said are inadequate for translational research.

In his C-SHALS talk, MD Anderson's Vogel noted that his organization has carried out a large-scale software development effort to create the informatics systems it needed. His staff developed ClinicStation, an electronic medical record; ResearchStation, which integrates research data with clinical information; and TissueStation, a tissue-bank-management platform.

Vogel said he and his colleagues turned toward software development more than ten years ago "largely because of shortcomings of commercial products in the clinical space."

Test results that include the expression of multiple genes associated with disease

progression, treatment response, or recurrence risk are very different from traditional lab test results with a single data point, which is what most hospital IT systems have been set up to handle, he said.

The University of Washington has also developed its own translational health system called MindScape. Peter Tarczy-Hornoch, a professor in the division of biomedical and health informatics at UW, said that when it was developed, there was no commercial product that could "get all the data in one place."

Tarczy-Hornoch spoke to *BioInform* last month at the American Medical Informatics Association's Translational Bioinformatics meeting, which he chaired.

The system's architecture allows integration of genomic information with clinical data, but he admitted that it is not an "optimal" system for all genomic medicine needs. He said that his group is continually developing the system and integrating it with vendor tools.

Comprehensive platforms require clinical data repositories that include all patient data, which is not the same as an electronic medical record, he emphasized. Lab systems hold data that is not present in a record, such as pathology reports and EKG system reports, which have many data elements.

Oftentimes, he said, "you are taking things from the source electronic system that exists as discrete data and it gets condensed down to text by the time it gets to the medical record system." For example, an automated EKG system report might be converted into a PDF or an image in the medical record system, Tarczy-Hornoch said. As a result, "there is a loss of computable data" as this information finds its way into the electronic medical record.

As genomic medicine moves into the clinic, "electronic medical record vendors are going to have to begin to support clinical data repositories and suck in data from their competitors," as well as home-grown systems, but these tasks are currently not straightforward, he said.

Furthermore, he noted that as whole-genome sequencing moves within reach of patients, it will be even more important to ensure that health information systems can interpret this information as part of care — for example by analyzing which genes, pathways, and networks are relevant to a given disease.

Decision-support systems in medicine currently rely on very simple rules, such as "if creatinine is above 1, do not give this drug which is excreted by the kidney," Tarczy-Hornoch explained. But it is a challenge to develop systems around complex information such as, "If you have this mutation and this mutation that is analyzed in this fashion," he said.

In addition to being able to interpret multiple variables, these new rules might need to incorporate information about a lab instrument, specimen handling, or automation techniques, he said.

Electronic systems will need to deliver "actionable" results and provide decision support for physicians in order to gain market acceptance, Joyce Mitchell, associate vice president for health sciences information technology at the University of Utah, told *BioInform* at the

AMIA meeting.

Physicians will likely be cautious about integrating genomic findings into their clinical decision-making, she said. As a result, decision-support systems in hospitals and physicians' offices must be able to handle and present complex data, which can include statistical analysis of SNP data or statistics-based risk assessment.

Genomeweb system

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