Newsbytes

Nebraska informaticians mine and translate genomic data

September 2018—More than five years have elapsed since clinicians at the University of Nebraska Medical Center approached the institution's informatics department with a problem. They wanted to more easily access structured genomic data stored in the EHR system for the diagnosis of cancer patients.

UNMC's Scott Campbell, PhD, director of informatics for the public health and pathology laboratories, and James Campbell, MD, professor of internal medicine, pondered the problem and set to work preparing a solution for coding and mining molecular pathology test results.

The complex genomic data created from sequencing pipelines are not represented in a form that is readable in EHRs, says Dr. Scott Campbell. Furthermore, raw genomic data are difficult to translate into actionable information that clinicians can use at the point of care. So, together, the doctors engineered a bioinformatic process to extract discrete molecular data—or "computable anatomic and molecular pathology facts," as Dr. James Campbell refers to them—from sequencing pipelines and convert them into a format that EHRs and clinical data warehouses can readily consume and that conforms to standards that EHRs support, so they can be disseminated broadly.

Because both men had experience with SNOMED CT (Systematized Nomenclature of Medicine Clinical Terms) and LOINC (Logical Observation Identifiers Names and Codes), with Dr. James Campbell having devoted more than three decades to developing SNOMED, it seemed natural to use these standard terminologies for their project. The issue, however, was that neither terminology could properly describe the molecular data, so the doctors worked with SNOMED International to develop extensions to the relevant codes and code definitions. "We authored those term extensions in conjunction with advice from our colleagues in the CAP Informatics Committee and with other molecular pathologists abroad as we tried to develop consensus on how to represent these data," Dr. Scott Campbell explains.

After receiving input from the CAP Informatics Committee, Drs. Scott Campbell and James Campbell created a question in SNOMED CT to address genetic variance at a particular gene locus. The answer for the variance that was observed was rendered in Human Genome Variance Society nomenclature. "We took that and embedded it into our molecular pathology software system, and our reports, upon sign-out, transfer that data, as HL7 messages, to our NebraskaCARES biobank software, and we also get a fully written report in PDF," says Dr. Scott Campbell. "The HL7 format is something that can be readily accepted by our Epic EHR," he adds, "and we are ready to begin steps toward implementation."

The PDF format, Dr. Scott Campbell points out, "is very helpful the first time the doctor reads the report. However, the PDF report gets filed away in our EHR system and is no longer easily found and the data in it is not computable." The computable data transferred to the EHR, on the other hand, can be presented as a flowsheet report that follows the patient longitudinally. That report can be presented in the EHR "almost as you would present a CBC," he says. "So when the physician is looking for a particular molecular result or is trying to determine if a certain test was or was not done yet, it's much easier to find in a flowsheet than it is to find looking through PDF documents. When a patient is no longer responding to a particular therapy, for example, this information is usually sought. It also enables physicians to surveil the patient."

The CAP recommends that molecular labs be able to surveil their results and observations to recall or identify patients with a particular variance, Dr. Scott Campbell continues. "As it relates to lab quality, this allows laboratorians to look at the molecular data and compare it to what they believe should be present or should be the norm for the lab. Are we over- or undercalling a particular number of variances that we would expect because there is a change in our environment? Is that because there is a change in our pipeline—in its performance? Has something fallen out of calibration or has the process altered? It's definitely something that the laboratorian can use to determine or assess the performance of their equipment if they see abnormal trends in the results they

produce."

All of these efforts feed into the goal of having "an intelligent EHR that will support cancer precision medicine," says Dr. James Campbell. "People are very excited that this is possible," Dr. Scott Campbell adds.

UNMC has published the terminology for the methodology "and it is currently being promoted into international standards," Dr. James Campbell notes. The terminology is accessible through the University of Nebraska website (www.unmc.edu/pathology/informatics/tdc.html) to those with a SNOMED CT license through the National Library of Medicine. Vendors that license the methodology can integrate the open-source terminology with their products.

The SNOMED community has reacted enthusiastically to this offering, says Dr. Scott Campbell, and feedback from molecular pathologists, oncologists, and other clinicians, as well as researchers, will help refine it. "It's certainly not a perfect system," he adds. "It's revolutionary, but that doesn't mean it's not going to have hurdles." —*Iulia Filip*

Mount Sinai center addresses blockchain in health care

The Icahn School of Medicine at Mount Sinai has established the Center for Biomedical Blockchain Research. "The ambitious venture—the first of its kind at any academic medical center—will place Mount Sinai on the cutting edge of research that uses the technology, a distributed, decentralized secure database system originally developed for Bitcoin, to solve problems in health care and medical science," according to a Mount Sinai press release.

The center will operate as part of Mount Sinai's Institute for Next Generation Healthcare, which was founded in 2016 to advance such technologies and disciplines as artificial intelligence, robotics, genomic sequencing, wearable devices and sensors, and cloud computing.

"Our aim is to understand whether blockchain and associated technologies can be used to solve open problems in health care and biomedical research," says Noah Zimmerman, PhD, director of the institute's health data and design innovation center.

Mount Sinai researchers working at the center will be able to conduct scholarly evaluations of blockchain-enabled solutions and build and test their own prototypes within the Mount Sinai Health System. The center also plans to partner with health care and technology companies working on blockchain projects for clinical medicine and biomedical research.

"At Mount Sinai, we bring to the table deep expertise in biomedical data, machine learning, and data governance," says Dr. Zimmerman. "This experience will allow us to address many of the most promising uses for blockchain in biomedicine with the goal of improving health care delivery and reducing costs."

NIST offers guide to secure EHR data on mobile devices

The National Cybersecurity Center of Excellence at the National Institute of Standards and Technology recently released free guidelines on how to protect EHR information on mobile devices.

"Specifically, we show how security engineers and IT professionals, using commercially available and open-source tools and technologies that are consistent with cybersecurity standards, can help health care organizations that use mobile devices [smartphones and tablets] share patients' health records more securely. We use a layered security strategy to achieve these results," the practice guide states.

The publication, "Securing Electronic Health Records on Mobile Devices," is broken down into five volumes: an executive summary; approach, architecture, and security characteristics; how-to guides; standards and control mapping; and risk assessment and outcomes.

The guide includes an example solution to the security issue, which was developed by the National Cybersecurity Center of Excellence using commercially available products. However, the center does not endorse such products.

“The example solution is described in the ‘how-to’ guide, which provides organizations with detailed instructions to re-create it,” the publication states. “The NCCOE’s approach secures patient information when practitioners access it with mobile devices.”

The guide can be viewed or downloaded at www.bit.ly/NCCOE_guide.

HudsonAlpha develops genetics tool to tackle phenotypic data

A team of researchers at the HudsonAlpha Institute for Biotechnology has introduced PyxisMap, a tool for zeroing in on the genetic variants that may be responsible for a patient’s medical condition.

“PyxisMap collects the symptoms of a patient in the form of free text or ordered lists, then runs the extracted terms against a data structure containing relationships between these symptoms (or phenotype terms) and genes and variants in order to rank these genomic regions based on disease associations,” according to a press release from HudsonAlpha, a nonprofit genetics and genomics research institute in Huntsville, Ala.

The tool can be used alone or in tandem with software for studying the likely impact of genetic variants identified in patients, such as HudsonAlpha’s CODICEM software. “Integration of phenotypic data allows identification of variants specifically related to the patient’s symptoms,” Liz Worthey, PhD, a member of the team that developed PyxisMap and director of software development and informatics at HudsonAlpha, told CAP TODAY. “The tool incorporates information on disease genes from standard disease databases as well as integrating the most up-to-date phenotype-gene associations recently published in academic journals.

“By combining both the variant impact and the phenotype ranking tools, you can quickly hone in on the variants underlying an individual patient’s disease,” Dr. Worthey continues. When used together in a pilot study, PyxisMap and CODICEM placed the causal variant among the top 20 potential variants more than 90 percent of the time, she adds.

PyxisMap is currently available for research use only. The technology is being used in several research projects focused on identifying deleterious genetic variation in people with rare or undiagnosed diseases.

For more information about PyxisMap, email Dr. Worthey at lworthey@hudsonalpha.org.

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