



Bridging the Genomics-Health IT Gap for Precision Medicine

By Jody Ranck

The Reality of Precision Medicine

Since the White House launched its Precision Medicine initiative in January 2015 there has been a great deal of buzz about personalized or precision medicine and the future of healthcare. “Personalized medicine” is an older term and is gradually falling by the wayside as critics think that it denotes a focus on the individual whereas precision medicine is more focused on which treatments work best for patients with a specific genetic, lifestyle or environmental context. The latter is more appropriate in the context of digital health where the growth in wearables, mHealth and even Population Health Management have become part of precision medicine initiatives.

But once we settle upon a definition, what are the real challenges to making precision medicine a reality? Given the significant challenges associated with EHR implementations as well as interoperability challenges across a given community, how will healthcare begin to address an additional stakeholder whose datasets are much larger and bring these insights into the clinic in an actionable way?

There are very few, if any, EHR examples today that have the capability of integrating, in a systematic way, genetic data in a format that can be readily used for treatment and therapeutic practice. This raises a number of important questions on when precision medicine can become reality in the clinic and what kind of strategic roadmap can be put into place to address the health IT issues?

Genomic Data and EHR data

Precision medicine requires combining phenotype data with genotype in order to prescribe a custom therapeutic regimen for the patient. For many genetic conditions the clinician also needs a comprehensive medical history to fully understand the individual’s genetic makeup.

Most phenotypic data in the EHR is in free form and exists in multiple record systems making it difficult for clinicians to have an accurate picture of the patient's genotype. The Meaningful Use (MU) criteria are of little help in this regard. Stage 2 requires family history data for only 20% of patients and no discrete capture of patient's historical information.ⁱ

The challenge with genomics also rests in the more dynamic nature of a constantly developing science that generates new conditions based on recently discovered deletions or mutations in the genome. Genetic tests can evolve whereas most other tests measure biometric data points for a discrete point in time. The gap between terminologies used in EHRs and those in genetic medicine is also problematic.

Another important difference is that most lab test data found in current EHRs is relevant to a precise episodic moment in a patient's life course in contrast to genetic testing data that will follow the patient for their entire life.

Genomic Data, Analytics and EHRs

Virtually all EHR data is stored in data warehouses that are SQL or object-based. While adequate for EHR systems these data warehouses are not readily compatible for genomic data for a host of reasons including:

- Slow retrieval of the very large datasets commonly found with whole genome sequencing.ⁱⁱ
- Genomic data is highly variant and traditional data warehouse technology struggles to quickly process these variants.
- Standards for storing whole genome sequencing data are still a challenge as commonly adopted standards for such data across the industry remain weak.

Some vendors are looking to tackle this issue through adoption of newer database technologies such as Cassandra, Elastic Search and Hadoop, which will allow HCOs to combine genomic and other datasets (claims, EHR, etc.) for advanced CDS in support of precision medicine.

Today, most genomic data is now made available in the EHR as a textual laboratory report but this data is not available for CDS systems in the same format. Another issue is how to store raw genomic data or partial segments of pathological variants in databases so that they can be readily accessed by CDS.

As the cost of sequencing whole genomes has fallen the issue of storing this data outside of academic research centers is becoming an issue for clinics. One strategy is to only store the pathological variants of the data in the EHR but this becomes a problem when it is medically necessary to have the whole genome.

The terminologies used in genetics are problematic as well. The current system uses descriptions of chromosomal abnormalities and mutations, but in many cases these are not associated with clinical syndromes. Often patients need to be described in the record in terms of a mutation or deletion that a relative has but they do not have themselves. This makes the whole family history record format as it now stands in EHRs challenging for actually doing clinical genetics.ⁱⁱⁱ

EHR Implementations and Customization

The nature of most EHR implementations can pose serious challenges to integration with genetics data. It is quite common to find specific types of medical practices calling for customized builds of EHR systems to match their clinical preferences. The configuration of customized EHR databases has implications for how easily the data in EHRs can be integrated with genomic data. Sometimes certain populations can be lost depending on the nature of the integration which means that data governance processes will need to have the foresight to anticipate precision medicine needs and factor this into database strategies early on to avoid costly and time consuming integrations later on.

Another aspect of EHRs that will need to be addressed is the current capacity for storage of genetic variants. Many can only store a set number of variants for specific genetic tests. When we move to more whole genome datasets this presents a severely limited option for actually doing precision medicine because most variants will not be able to be stored.

Laboratory systems are another major challenge. Some of the larger EHR providers may integrate lab systems with EHRs but this is rare and now we have the genetic tests to add to the mix and the need to represent and integrate data from one system into another. Precision medicine is heavily dependent on lab tests that can help further calibrate treatments for individuals.

Clinical Decision Support Systems (CDS)

One of the challenges is the current rules-based systems used in EHRs for most CDS systems may not be that applicable for the types of data and conditions represented by genomic data or if they can scale for the granularity in mutations and deletions in large genome datasets. Some oncology practices are already using brief synopses of the most important genetic data for a given patient in the patient dashboard, for example. Genetic medicine experts are worried, however, that current approaches used in laboratories using LOINC and HL7 standards are not sufficient for linking genomic data to CDS platforms and genomic data are still not captured in a structured format that can easily be integrated.^{iv}

Bridging the Genomics-Health IT Gap

To address most of the challenges listed above a multi-center partnership was created to bridge the gap between genomics and health IT systems. The network includes Group Health Cooperative/University of Washington, Mayo Clinic, Vanderbilt University, Marshfield Clinic and Northwestern University. The Electronic Medical Records and Genomics (eMERGE) Network was founded with nine academic research centers that have their own bio-repositories that are linked to phenotypic data stored in EHRs.^v

An earlier initiative, CABig, by the National Cancer Institute that sought to build a bioinformatics grid to facilitate sharing of genomic data became a bureaucratic nightmare with substantial cost overruns and poorly designed technological systems., eMERGE seems to have escaped these challenges so far and is beginning to see some success in developing actual tools for clinicians and researchers.

One of the most successful areas is the development of pharmacogenomics tools for clinicians such as the warfarin sensitivity application. Another tool, the Infobutton Manager, helps link clinicians to online genomic resources. OpenCDS is the interface component that works with WarfarinDosing.org for personalized approaches to warfarin dosing.

Some participants in the eMERGE Network are associated with the SMART platforms project at Harvard are calling for an “Apps-Based Information Economy” that makes better use of open APIs for moving large data sets and extracting insights from this data more rapidly.^{vi} App-enabled EHRs could enable a much wider range of functionality on top of the EHRs as well as enable exchange of EHR data into research platforms for use in clinical and translational research. The developing HL7 FHIR standard could play a pivotal role here.

The Precision Medicine Initiative plans to enroll one million patients via collaborations with NIH and the Federally Qualified Health Centers through their Cohort Program. The question remains, however, as to how the latest advances in genomics will meld with existing health IT systems to support clinicians at point of care. The challenges are significant and will likely be tackled in piecemeal fashion at some of the largest research institutes and trickle down over years, if not a decade or more, into common practice.

ⁱ Keith Marsolo and Andrew Spooner. Clinical genomics in the world of the electronic health record. *Genetics in Medicine*, 15(10):786. October 2013.

ⁱⁱ Ury, Andrew. Storing and interpreting genomic information in widely deployed electronic health record systems. *Genetics in medicine*, 15(10): 780.

ⁱⁱⁱ Marsolo and Spooner, p. 789.

^{iv} Overby, Casey Lynnette et al. Opportunities for genomic clinical decision support interventions. *Genetics in Medicine*, 15(10):820. October 2013. And

^v Gottesman, O. et al. The eMERGE Network: past, present and future. *Genetics in Medicine*, 15(10): October 2013.

^{vi} Mandl, K., Mandel, J; and Isaac Kohane. Driving Innovation in Health Systems through an Apps-Based Information Economy. *Cell Systems*,1, July 29, 2015.

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