

The Real Opportunity of Precision Medicine and How to Not Miss Out

By David Crockett, PhD

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The President’s recent [comments on precision medicine](#) during the State of the Union address seem to have captured the attention of the healthcare world. What lasting impact this announcement, and its proposed \$215 million investment, will have on the healthcare industry is anyone’s guess. One thing is for certain—healthcare is changing!

Many observers feel that precision medicine is the magic panacea that will allow [health information technology](#) to leverage advances in genomics with emerging methods for managing and analyzing large data sets, and to accelerate research and biomedical discoveries—while still protecting patient privacy. But what exactly is precision medicine?

What Is Precision Medicine?

Medical treatments are often designed for the so-called “average patient.” This one-size-fits-all approach to therapy can be successful for some patients, but perhaps not for others. [A White House fact sheet](#) from January 2015 titled “President Obama’s Precision Medicine Initiative” defines precision medicine as, “a new model of patient-powered research that promises to accelerate biomedical discoveries and provide clinicians with new tools, knowledge, and therapies to select which treatments will work best for which patients.”

While some would argue that this personalized approach is not really all that new to medicine, we can all agree that tailoring medical treatment to the individual patient makes a lot of sense. To state this more simply, precision medicine understands each patient’s individual illness and delivers the right treatment at the right time.

Clinical improvements to [reduce system variation](#) in care delivery can lead to high-quality health systems with lower cost for patients. In contrast, precision medicine seeks to understand pertinent patient variation from an individual’s unique environments and biochemistries. So on one hand, reducing variation in healthcare is a good thing, but provision for patient variation in treatments is also necessary.

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Precision Medicine Starts With Precise Registries

Remarkable advances in DNA sequencing technology are leading to more and more disease discoveries. This, in turn, allows for more treatments tailored to specific characteristics of individuals receiving the care. This molecular transformation of medicine is currently most visible in areas such as cancer, where patients with breast, lung, and colorectal cancers now undergo genetic testing as part of routine patient care. This permits the treating physicians to select a therapy ahead of time that will improve a patient's chances of survival and reduce exposure to adverse effects. In essence, understanding the genetic variation removes the guesswork of choosing an effective therapy.

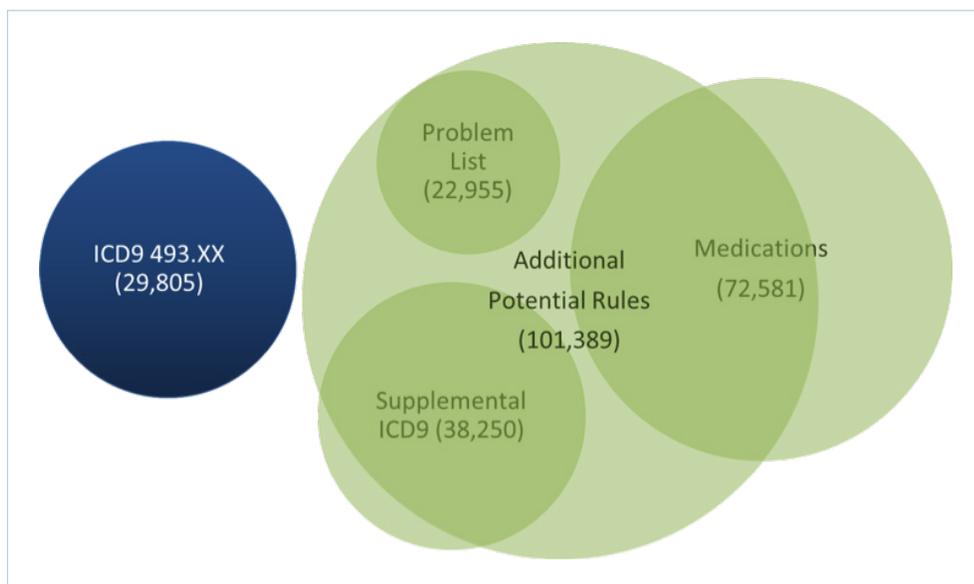


Figure 1: Precise patient registries go far beyond simple inclusion/exclusion lists of ICD codes.

The key to leveraging this patient variation is precise registries. For example, instead of ICD-code based inclusion only, a more comprehensive registry would include keywords from problem lists, patients with medications specific to that condition, and other types of supplemental criteria. The difference in patient counts can often be pretty drastic as shown in Figure 1, where relying on the ICD-9 diagnosis code for asthma alone (n=29,805) compared to incorporating related terms and medications, yields a far more complete picture of patients and disease (n=101,389).

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Without precise definitions and registries of patients and disease, you can't have precise clinical research, precise financial and risk management, personalized (precise) healthcare, or predictable clinical outcomes. Moving into the future, this becomes increasingly important for gene test results (and associated interpretations) to smoothly integrate with the EHR and drive actionable medical decisions.

Precise Medical Decision Making

We live in a remarkable era of information, when all that is known about a person—from family history and genetics to location history and environment—can be balanced against all that is known in the medical domain. In short, this big-picture view of medical decision making can allow care providers to focus both prevention and intervention techniques on appropriate individuals, while avoiding unnecessary cost or unwanted side effects for those patients who would not benefit. (See Luke Timmerman's post [What's in a Name? A Lot. When It Comes to "Precision Medicine."](#))

Traditional laboratory testing for point mutations and single gene assays are now being replaced by high-throughput gene panels, and exome or genome sequencing. Delivering gene test results, associated interpretation of gene variants, and appropriate clinical action in a timely manner becomes an important key to realizing the promise of precision medicine. (My colleagues and I published a study on this in the May 2012 edition of *Genome Medicine* called [Consensus: a framework for evaluation of uncertain gene variants in laboratory test reporting.](#))

Mainstream EHR and health data warehouse offerings in the U.S. have begun to focus on capturing this genomic market segment. They are pushing toward the end-goal of sequence variant information coupled to clinical utility—but many lack a convenient tie from the rapidly advancing sequencing platforms and bioinformatics pipelines to the existing clinical and EHR environment.

Many experts liken this integration of genomics into the EHR to [mapping "terra incognita."](#) In fact, an entire issue of [Genetics in Medicine](#) was recently devoted to this very topic. Unfortunately, the ability to practice precision medicine is largely dependent on gene variant knowledge bases, standard meta-data architecture, and decision-rule engines being made available to assist clinicians when taking action based on test results. In this important effort, the ability to link knowledge among databases on genotype, phenotype, and clinical outcomes is crucial. For more information on this, see the September 2014 edition of *Pharmacogenomics and Personalized*

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Medicine for a study by Huser, Sincan, and Cimino entitled [Developing genomic knowledge bases and databases to support clinical management: current perspectives.](#)

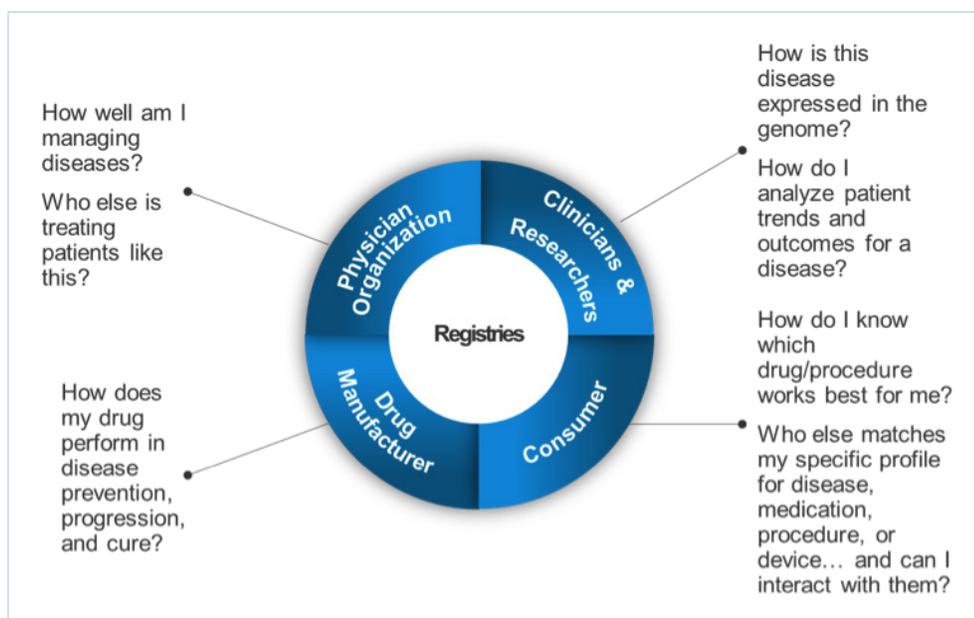


Figure 2: Key questions that surround the data and technology of precise patient registries.

As shown in Figure 2, this integration of research data, clinical data, drug data, and patient data is the original motivation for building disease or patient registries. In other words, a precise registry can provide specific answers to specific audiences—even when the questions may not be anticipated ahead of time.

In short, precise registries drive precise medicine. 🗨️



David K. Crockett, Ph.D. is the Senior Director of Research and Predictive Analytics. He brings nearly 20 years of translational research experience in pathology, laboratory and clinical diagnostics. His recent work includes patents in computer prediction models for phenotype effect of uncertain gene variants. Dr. Crockett has published more than 50 peer-reviewed journal articles in areas such as bioinformatics, biomarker discovery, immunology, molecular oncology, genomics and proteomics. He holds a BA in molecular biology from Brigham Young University, and a Ph.D. in biomedical informatics from the University of Utah, recognized as one of the top training programs for informatics in the world. Dr. Crockett builds on Health Catalyst’s ability to predict patient health outcomes and enable the next level of prescriptive analytics – the science of determining the most effective interventions to maintain health.