Published on *American Council on Science and Health* (https://www.acsh.org)

Precision Medicine Stands On Imprecise Infrastructure

By Krystal Alexander — August 23, 2016

Credit: Shutterstock [1]

What is precision medicine?

Precision medicine stands to be the future of healthcare. A future painted as delivering personalized medicine and targeted therapies, doing away with the one-size-fits-all approach and exploiting the root cause of a disease to find curative/therapeutic options. It's a beautiful, grand concept, that the current medical and scientific world's infrastructure is simply not prepared for to be more than a concept.

Precision medicine hinges largely on genomics – the study of our genes and their function. This relatively new field grew out of the human genome mapping project. Genomic medicine employs molecular biological techniques to identify a gene, gene variants or protein that may be the cause of a disease. Once located and identified, researchers work to develop a treatment plan that targets or modifies the gene or it's protein, customizing or personalizing care.

To be successful genomic medicine requires basic science and medical science to work hand in hand. And it also requires an integrated, robust information system to analyze genomes and allow the information to be utilized by healthcare practitioners.

Barriers to precision medicine

Undoubtedly genome mapping has proven to be helpful for patients. Their stories are the ones highlighted by the media as miraculous answers to debilitating disorders. They are documented in scientific publications by practicing physicians as case report studies. And they are just that — individual cases. We have seen how genomics can help the individual. But what about the whole?

Why haven't we been able to reproduce this for thousands of other patients who have been unresponsive to conventional treatment?

For some diseases, it goes back to basics. Understanding the molecular mechanisms is crucial. There are drugs on the market that are effective treatments for patients, but how or why they work remain elusive. Precision medicine takes the opposite approach, attempting to create a drug with a well defined mechanism of action, based on the root of the disease.

- **Electronic Medical Records**

A genomic dataset is extensive and requires a technologically efficient system that stores, analyzes and easily transmits data to those authorized to access the data. Many critics argue that the electronic medical record system is not currently able to facilitate the basic infrastructure necessary for genomic data use in real-time in a clinic or hospital setting. Stanford Medicine and Google [2] are collaborating to change this – building cloud based applications capable of securely storing, processing, analyzing and sharing the massive genomic data sets.

- **Lack of Diversity**

One of the goals of genomic studies is to create a genetic database accounting for every ethnicity. To date, many genetic studies have focused on Caucasian populations, which may not be applicable to other racial groups. Diversity in genomic medicine allows characterization of underrepresented populations, possibly providing explanations as to why some are more susceptible to certain diseases or why the rate of progression of a disease in a particular ethnicity is comparably faster.

- **Physician Education**

Genomics became a specialized area of medicine as a result of the human genome mapping project, a testament to the constantly evolving nature of medicine. But many of our current healthcare practitioners didn't have the opportunity to learn these concepts during their medical training, creating a generational knowledge gap. Doctors need to be introduced to the field and its applications in the provision of healthcare.

Primary care doctors rely on evidence based medicine to treat patients. But most genomic tests haven't been through rigorous testing or had the longitudinal studies assessing their effects on patient outcomes. Genomics is still in its infancy and the benefits versus risks have yet to be fully established. These reasons contribute to the hesitancy to incorporate genomic testing into primary care practice.

- **Patient Education**

Increasing numbers of patients seek genetic testing. And what makes this dangerous is that with the advent of companies like 23andMe, anyone can sign away the rights to their DNA to get a genetic story that has no impact for their current or future health. Despite their attempts to comply with FDA restrictions that state these tests should not be utilized to diagnose medical conditions, companies like 23andMe thrive on the curiosity, naivety and the genetic illiteracy of patients. These patients then approach their physicians demanding this information to be put into the

context of their healthcare, essentially seeking medical advice after the fact. While the patient may believe they are being proactive about their health, this isn't always the case. Sometimes they are just being a financially easy target for exploitation.

- **Finances and Accessibility**

That brings us to the high cost of genome sequencing. Though researchers have found ways around this, using targeted testing for specific genes. But cost raises the issue of accessibility. The implied goal of personalized medicine is to make genomic testing available to all regardless of socioeconomic status. How are we going to take something so costly, and make it available to the masses?

As it is with most things in life, it's easy to spot the problems, but not the solutions. The goal of precision medicine is admirable and certainly not elusive. However, it will require initiatives and collaborations among various disciplines, and an integrated network infrastructure built on a secure foundation.
