Personalized Medicine Can't Wait for Genomic Data

written by Robert McNutt, M.D. | August 10, 2016

Personalized medicine is off and running. The effort to enroll one million people who will volunteer their genomes for science by the year 2019 kicked off recently with an event featuring President Obama that included more than 150 of the first volunteers.

But this effort is not for my patient. It will be either too little, or too late, and certainly not enough.

While personalized medicine is an old concept, the new push for personalization focuses on genes or gene products. These, it is hoped, may be better predictors of an individual's outcome of a disease condition. The new efforts may even redefine some disease classifications, as well as further subdivide people by their predicted outcomes. The key word here is "further."

The science of medicine has been personalizing care since inception. Genes offer a new target for personalization, but not a new concept. But, until this dream is either realized or dashed, we must do better with present technologies.

Personalized Medicine is About the Variation—Not the Average

I am a decision-making consultant. The recent experience of a person in my care highlights the need for a better understanding of individual variations, but at the same time, shows how the concept of personalized care is presently poorly defined and prematurely communicated.
Those involved in the research or selling of personalized products define personalized medicine differently, but at the core of the definition is a "subdivision of individuals from the group."
Personalized medicine is about the variation and not the average.
My patient is a young woman with breast cancer, offered treatment for her cancer based on an

“old system ” of subdividing the variations in women with cancer, size of tumor, certain tumor characteristics and node status. She was presented a plan that many, if not all women, in her subdivision are offered. Yet, her diagnosis comes in the transition period between old knowledge about personalization and new. She has an unusual biology tumor. In fact, so unusual that she has a lower chance than any of the other subdivided individuals to derive benefit from the proposed, onerous treatment plan.

The woman actually had data from a randomized clinical trial (RCT) available for her decision-making. This is, according to present day theories, a plus. RCT data is supposed to be the best. However, if it is best, and I don’t think so—at least as presently conducted—it is not best for individuals who must decide. This is because the RCT is a technology of the average, and my patient, and yours, is anything but average.

Personalized Medicine Requires Better Research

The RCT pertinent to her care captured information about some subdivisions such as receptor status, but in any subdivided group, there were few patients. This was especially true for her. She has Factor A *and* Factor B *and* Factor C. For her, she was a group of the combined probabilities of each. She was a small subgroup patient. In that subgroup, the confidence estimates ranged widely and pointed to little benefit from her proposed plan.

She was stuck. I could not tell her for certain what her personalized outcome portfolio might be. But, I showed her all the data, showed the average difference in outcomes and the variation about the average, showed her that the study only went on for a brief follow-up time of four years, showed her the variation in how her specific tumor type did in the study, showed her the actual number of patients who suffered outcomes in compared groups, showed her that the study did not even subdivide accurately her tumor type, showed her the table with side effects and then discussed how studies can be wrong and how they can be right. Three hours of discussion, three hours of highlighted uncertainty, three, for her, gut-wrenching hours of unease.

While we wait for genes to restructure the practice of medicine, we also must do a better job of doing studies for my patient. We have discussed clinical data repositories on full populations of patients in other blogs and have claimed superiority of research designs from such complete and universal data collection efforts.

Personalized Medicine Will Depend on Specific Disease Condition Clinical Repositories

A complaint has been, “What do we do with diseases and individuals that are low in prevalence in any single clinical repository?” Well, we build specific disease condition clinical repositories

that include all patient data on subdivisions known and presumed (saving materials if we must). This may seem a tough task, but is simple, actually. It is just data collection and would take planning, certainly.

However, if we can gather a million people for gene studies, can't we gather thousands of useful data elements from clinical data repositories? Without such planning and organized registries, individuals will be doomed as decision makers. In fact, the RCT is incompatible with personalized care, as the technology does not adequately subdivide. The RCT left my patient unsatisfied once she understood the uncertainties in the study. There has to be better ways to study individuals.

I hope, only for the purpose of better data, that among the one million people joining for genome science with breast cancer, there are some with my patient's problem. Even if a handful do, then we can gather them together and study to see if that subdivision is worth anything to them. Personalized medicine will need research designs as innovative and futuristic as the genomic results. There is much to do to reach the potential of new tools of personalized care. These tools are perhaps better than old, but only studies will tell us.

Until then, the only definition of personalized care should be synonymous with informed care. Patients must be given the information with all of the "warts." They will know what to do with the information better than we will. Let us not forget, the best personalization agent is the patient.

By the way, my patient decided not to take her physician's proposed aggressive treatment plan, but chose, instead, one targeted to her biology. She told me that the lack of personalized information gave her hope that doing less may be better for her, especially traded off against the side effects. That sounds like personalized decision making to me.

Image Credit: Jin Neoh