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POLICY

Toward Precision Medicine: A New Social Contract?

EARLIER THIS YEAR, ILLUMINA INC. AND LIFE TECHNOLOGIES INC. EACH ANNOUNCED new products that can sequence a genome for \$1,000 in a single day (1); approximately 3 million times cheaper than the cost during the Human Genome Project back in the early part of the last decade. Furthermore, cloud-based, big-data software companies are capable of using whole- and partial-genome sequencing to automate and operationalize diagnostics in real-life situations with patients. But no one believes that less expensive data and more analyses are in themselves enough to accelerate the path to disease cures. In fact, science writer Nicholas Wade asserted in the 12 June 2010 issue of the *New York Times* that, a decade after completion of the first draft of the human genome sequence, too little clinical benefit has been realized (2). Many have subsequently defended the human genome project's benefits; but it remains reasonable to ask why, given the explosion of scientific knowledge in the last decade, haven't we seen greater gains in health outcomes?

A new National Research Council report (3) from the U.S. National Academies (<http://www.nas.edu/nrc>) attempts to address this question. The report calls for a new data network that integrates emerging research on the molecular makeup of diseases with clinical data from individual patients to drive the development of a more accurate classification, or taxonomy, of disease that ultimately enhances diagnosis and treatment. The new taxonomy of disease integrates multiparameter molecular data with clinical data (including the traditional physical signs and symptoms), environmental data, and health outcomes in an iterative fashion. The ultimate goal is the devising of a kind of "Google map for health" that would be both dynamic and flexible. The committee coined the term "precision medicine" to refer to how the new taxonomy could allow for the tailoring of medical treatments to the individual characteristics of each patient, using "precision" in the colloquial sense to mean both "accurate" and "precise."

Imagine a world in which the vision outlined in the National Research Council report became a reality. The new data network could improve biomedical research by enabling scientists to access patients' information during treatments while still protecting the patients' rights. This ability would allow the marriage of molecular research and clinical data at the point of care, as opposed to research information continuing to reside primarily in research labs or publications. Data would be continuously deposited by the research community and extracted directly from the medical records of participating patients. Having this access integrated with clinical care could reduce the cost of research, make scientific advances relevant to real-life medicine, and facilitate the use of electronic health records (4). As a physician who has worked in the pharmaceutical industry, this vision certainly resonates with me—in particular, the potential to more efficiently discover and develop transformative medicines such as trastuzumab for HER2-linked breast cancer or vemurafenib for BRAFV600E-mutated melanoma.

PATIENTS AND FORTITUDE

Anyone engaged in the currently daunting task of disease prevention, diagnosis, or therapy is immediately excited by this vision, but major impediments exist. Two of the most daunting bottlenecks are the low rates of participation in clinical trials and the current emphasis on patient privacy in research and clinical settings. After all, fewer than 5% of patients currently participate in clinical trials, and the Health Insurance Portability and Accountability Act (HIPAA) promises penalties for any who breach patient privacy, resulting in ever-increasing conservatism regarding access to and pooling of data.

I believe that the most important requirement for the new knowledge network envisaged by the Precision Medicine report is that it be driven by patients. Indeed, it is patients who uniquely understand the potential value of a social contract in which patients both contribute personal clinical data and benefit from the knowledge gained through the collaboration. Patients are also in the best position to demand the sharing of both data and professional credit that will be necessary to fully capture the value of this new collaborative approach to

acquiring, synthesizing, and widely disseminating biomedical knowledge (5). Patient advocacy can best ensure that policymakers in the U.S. Congress and elsewhere understand that well-intended efforts to guard patient privacy could impede the kind of data sharing required to accelerate the cures all are awaiting.

This opportunity to create an entirely new way to classify—and therefore understand and treat—human disease could bring us to the tipping point at which the remarkable scientific advances in biomedicine and engineering translate to concrete therapeutic benefits for humankind. We need only look back to the human immunodeficiency virus (HIV)–AIDS epidemic during the 1980’s to experience the power of patient advocacy combined with the dogged pursuit of scientific discovery and translation; clearly, motivated patients and scientists as well as their advocates can influence political, scientific, and regulatory agendas to drive advances in health.

Many of today’s most pressing health challenges may seem much less dramatic than the HIV/AIDS epidemic of the 1980’s, but the vision outlined in the Precision Medicine report (3) is worthy of that kind of passion and leadership. This vision is also an opportunity for a unified push by patient advocates on behalf of all patients, a movement that potentially can combat the inefficiency of competition between multiple disease-specific groups, as pointed out by Amy Dockser Marcus in the *Wall Street Journal* in 2006 (6). A unified group of patient advocates pushing government, academia, private industry, and caregivers to create a new social contract in which patients both contribute and benefit would be a powerful force. I cannot imagine a more effective way to create the world we imagined in “Precision Medicine” a reality.

- Susan Desmond-Hellmann

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