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ADVANCING KNOWLEDGE, PROTECTING INFORMATION: PRECISION MEDICINE AND POPULATION HEALTH RESEARCH IN HEALTH SYSTEMS

*Raj C. Shah, Alissa Bugh, Mary Jane Welch, Andrew Reeder**

I. INTRODUCTION

In order to achieve the highest value for the resources spent on the provision of health care, the delivery of health care in the United States must change. The health care enterprise in the United States strives to reclaim global leadership in achieving better health care outcomes for a diverse and inclusive population, better care experiences for individuals, and better use of resources for health and care. Achieving these goals requires the transformation of the health care system into a learning health system where practice influences research and research influences practice. Linking research with practice requires understanding the health of large groups of individuals as well as understanding the health of single individuals – an idea that is often described simply as “precision medicine.” Forces accelerating precision medicine include patient partnerships, mobile technologies, genomics, data science, and electronic health records. Forces resisting change include various laws created at times when technological advances did not exist. In this article, we explore the tensions between technologies advancing large scale health data sharing and health data privacy and security. We highlight potential evolutionary changes to help reconcile the tensions along with more disruptive innovations.

II. BACKGROUND

Why Health Care Must Change

Health care in the United States needs to undergo significant changes. In the last almost 80 years, the life expectancy at birth for women has increased from 65.2 years in 1940 to 80.1 years in 2003.¹ For men, the

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¹ Donna L. Hoyert, et al., *Deaths: Final Data for 2003*, 54(13) NAT’L VITAL STAT. REP. 1, 6 (2006), https://www.cdc.gov/nchs/data/nvsr/nvsr54/nvsr54_13.pdf.

life expectancy at birth has moved from 60.8 years in 1940 to 74.8 years in 2003.² The total number of persons over age 65 has increased from 9 million in 1940 to 47.8 million in 2015 and is projected to increase to 98.2 million by 2060.³ While life expectancy has increased, persistent gaps in health equity continue to be present even though reduced. For instance, the difference in life expectancy at birth between black men and white men was slightly above 7 years in 1970 but was still 6.3 years in 2003.⁴ The difference in life expectancy at birth between women and men was 4.4 years in 1940 but had increased to 5.3 years by 2003.⁵ In a recent report by the Commonwealth Fund comparing health care systems on dimensions of quality care, access, efficiency, equity, and healthy lives, the United States far exceeded the remaining 10 developed nations with a health expenditure per capita (in 2011 US dollars) of over \$8,500 while the second closest nation (the Netherlands) spent a little over \$5,600.⁶ For that expenditure of resources, the United States received an overall health care ranking of 11 while the United Kingdom received a ranking of 1.⁷ The highest ranking the United States received was 3 out of 11 for effective care.⁸ These issues have led the United States over the last 5 years to embark on achieving the “Triple Aim,” which includes: (1) better outcomes for the local population, (2) better care and experience for individuals, and (3) better use of resources for health and care.⁹

How Can Health Care Change

In order to achieve the Triple Aim, health care must be transformed into a learning health system.¹⁰ In a learning health care system, practice influences research and research influences practice. A continuous cycle is needed for learning health systems to engage with their environments, adapt and find solutions. Internal and external scans are required to identify problems. With the identification of problems that matter, design of a solution plan is needed that can be implemented in pilot settings. Collecting data and analyzing data can evaluate what works or does not work. The evidence generated leads to solution optimization in an iterative process. Finally, solutions are shared broadly to improve care for everyone through informing policy. The idea of a learning health system has been incorporated into a cyclical model of the United States Food and Drug Administration (“FDA”) to optimize the cycle of driving discovery science to early translational steps followed by clinical trials, clinical practice guidelines, performance measures, and outcomes.¹¹

² *Id.*

³ DEP’T OF HEALTH & HUMAN SERVS., *A Profile of Older Americans: 2016*, [https://www.acl.gov/sites/default/files/Aging and Disability in America/2016-Profile.pdf](https://www.acl.gov/sites/default/files/Aging%20and%20Disability%20in%20America/2016-Profile.pdf).

⁴ Hoyert, *supra* note 1, at 26.

⁵ *Id.* at 5.

⁶ Karen Davis et al., *Mirror, Mirror on the Wall, 2014 Update: How the U.S. Health Care System Compares Internationally*, THE COMMONWEALTH FUND (JUNE 2014), <http://www.commonwealthfund.org/publications/fund-reports/2014/jun/mirror-mirror>.

⁷ *Id.*

⁸ *Id.*

⁹ Donald M. Berwick et al., *The Triple Aim: Care, Health, and Cost*, 27(3) HEALTH AFFAIRS 759-69 (2008).

¹⁰ Sarah M. Greene et al., *Implementing the Learning Health System: From Concept to Action*, 157(3) ANN. INTERN. MED. 207-10 (2012).

¹¹ Rachel E. Sherman & Robert M. Califf, *What We Mean When We Talk About EvGen Part II: Building Out a National System for Evidence Generation*, U.S. FOOD & DRUG ADMIN. (May 3, 2016), <https://blogs.fda.gov/fdavoic/index.php/2016/05/what-we-mean-when-we-talk-about-evgen-part-ii-building-out-a-national-system-for-evidence-generation/>.

Why Population Health Research

Most research to understand health and health care delivery focuses on information collected from sub-groups. Results from limited samples then are extrapolated to the entire population using statistical techniques. As a result, there is a higher chance for research to have external validity as the sample selected may never fully represent the entire population. Cost of data collection is a large contribution to needing sampling techniques in research. The closest data collection mechanism to engage every potential person in the United States is the US Census. Written into the US Constitution and required to be conducted every 10 years, the attempt to count every single person in the United States had a cost of approximately \$12 billion in 2010.¹² However, the cost of using the US Census infrastructure even on an annual basis is dwarfed the estimated \$3.2 trillion spent by the US in health care in 2015.¹³ In addition, recent advances in science and technology are now accelerating the ability to potentially understand larger populations in more detail using very cost-efficient tools. These advances are tempered by forces opposing the growth of population health research.

Forces Accelerating Change

Forces accelerating change include, but are not limited to, genomics, electronic health records, and data sciences.

Gregor Mendel's 1865 presentation on the laws governing the transmission of heritable traits in peas is commonly viewed as the foundation of genetics as a scientific discipline.¹⁴ However, it took almost a century to determine that DNA was the biologic structure governing inheritance.¹⁵ Further foundational work led to the willingness to initiate the Human Genome Project in 1990 to characterize the DNA in human chromosomes, including all genes. It took until 2003 for the human genome sequence to be completed.¹⁶ In 2003, the cost associated with generating the first complete human genome sequence was estimated to be between \$500 million and \$1 billion dollars.¹⁷ With advances in technique in the last decade, the cost of sequencing the 23 chromosomes and 3 billion base-pairs of the human genome has dropped to about \$1000 dollars.¹⁸ Currently,

¹² U.S. DEP'T COM. OFF. INSPECTOR GEN., *Census 2010: Final Report to Congress*, Final Report No. OIG-11-030-I 1, 2 (June 27, 2011), <https://www.oig.doc.gov/OIGPublications/OIG-11-030-I.pdf>.

¹³ CTR. FOR MEDICARE & MEDICAID SERVICES, *National Health Expenditures 2016 Highlights*, <https://www.cms.gov/research-statistics-data-and-systems/statistics-trends-and-reports/nationalhealthexpenddata/downloads/highlights.pdf>.

¹⁴ NAT'L HUM. GENOME RES. INST., *Timeline: From Darwin and Mendel to the Human Genome Project*, <https://www.genome.gov/25019887/online-education-kit-timeline-from-darwin-and-mendel-to-the-human-genome-project/> (last updated Mar. 5, 2016).

¹⁵ *Id.*

¹⁶ *Id.*

¹⁷ NAT'L HUM. GENOME RES. INST., *The Cost of Sequencing a Human Genome*, <https://www.genome.gov/sequencingcosts/> (last updated July 6, 2016).

¹⁸ *Id.*

every person is able to submit a DNA sample to a commercial entity and for about \$200 obtain their ancestry and basic health information.¹⁹

In 2010, efforts significantly increased to digitize health care records. In 2008, there were no states where the percent of non-federal acute care hospitals that had adopted at least a Basic Electronic Health Record was over 60%.²⁰ By 2014, all States had more than 60% of their non-federal acute care hospitals with adoption of a basic electronic health record.²¹

The explosion of health data from multiple electronic sources along with sophisticated data science tools to organize and analyze the data has led to the development of highly dimensional information. Within a single person, we are able to understand various levels of information from the clinical data from medical histories, the genome, the transcriptome, the proteome, the metabolome, and the microbiome.²² While the genome represents an organism's complete set of DNA including all of its genes, the transcriptome is the set of all messenger RNA (the molecules that translate genes into proteins) in a single cell or population of cells and the proteome is the entire set of proteins expressed by a genome in a given type of cell at a given time under defined conditions. In addition to the detailed understanding of the pathways connecting DNA to proteins, the metabolome refers to the complete set of small-molecule chemicals found within a biological sample and the microbiome is the curation of microorganisms and their collective genetic material present in or on the human body. Given that advanced tools and technology related to "-omics" are not able to process a large number of samples in semi-automated or automated ways at a reduced expense, this level of detail on large numbers of individuals can be collected effectively. Using data science and computing technologies, we can analyze the network of relationships between these multiple layers along with relationships with community level and environmental data sets.

These advances, in addition to mobile technologies and patient partnerships, have led to the National Institutes of Health proposing the Precision Medicine Initiative to better appreciate how the connected, multi-level information can be utilized.²³ Precision medicine is an emerging approach for prevention and treatment of disease that takes into account people's individual variations in genes, environment, and lifestyle.²⁴ The scientific evidence to move the precision medicine concept into clinical practice is underway. The All of Us Research Program is one component of the Precision Medicine Initiative.²⁵ It involves engaging over 1 million volunteers residing in the United States who will share genetic data, biological samples, and diet/lifestyle

¹⁹ Websites such as 23andme.com and www.ancestry.com provide services to ascertain a person's ancestry.

²⁰ JaWanna Henry et al., *Adoption of Electronic Health Record Systems among U.S. Non-Federal Acute Care Hospitals: 2008-2015*, OFF. NAT'L COORDINATOR FOR HEALTH INFO. TECH. (May 2016), <https://dashboard.healthit.gov/evaluations/data-briefs/non-federal-acute-care-hospital-ehr-adoption-2008-2015.php#figure1>.

²¹ *Id.*

²² Marylyn D. Ritchie et al., *Methods of integrating data to uncover genotype-phenotype interactions*, 16(2) *Nature Reviews Genetics* 85-97 (2015).

²³ Francis S. Collins & Harold Varmus, *A New Initiative on Precision Medicine*, 372(9) *N ENGL J MED*. 793-95 (2015).

²⁴ NIH U.S. NAT'L LIBR. MED., *What is precision medicine?* (Dec. 19, 2017), <https://ghr.nlm.nih.gov/primer/precisionmedicine/definition>.

²⁵ NAT'L INSTITUTES HEALTH, *PMI Cohort Program announces new name: the All of Us Research Program* (Oct. 12, 2016), <https://www.nih.gov/all-of-us-research-program/pmi-cohort-program-announces-new-name-all-us-research-program>.

information that can be linked with their electronic health records, if they choose.²⁶ In this new model of doing science through engaged participants, responsible data sharing, and privacy protection, the generalizable knowledge from the cohort will (1) advance pharmacogenomics (the right drug for the right patient at the right dose), (2) identify new targets for treatment and prevention, (3) test whether mobile devices can encourage healthy behaviors, and (4) lay a scientific foundation for precision medicine for many diseases, common and rare.²⁷ The National Evidence Generation Infrastructure by the United States FDA also is being proposed to link data from multiple sources with the National Institutes of Health (“NIH”), the Patient-Centered Outcomes Research Institute (“PCORI”), the Centers for Disease Control and Prevention (“CDC”), and industry to impact medical product safety surveillance.²⁸

Forces Resisting Change

In the movement towards greater health data utilization for research, forces resisting change include various laws created at times when technological advances did not exist. Before the Health Insurance Portability and Accountability Act (“HIPAA”) was signed into law in 1996, there was not a patient privacy standard at the federal level for Protected Health Information.²⁹ HIPAA simplified the administrative processes for health data transfer for health care treatment, operations, and payments and provided administrative, physical and technical safeguards for Protected Health Information.³⁰ Through the passage of the American Recovery and Reinvestment Act – Health Information Technology for Economic and Clinical Health (“HITECH”), the HIPAA Privacy Rule was extended to include new civil penalties for violations, to have covered entities and business associated comply, and to establish breach notification obligations in an Act mainly designed to provide specific incentives to accelerate the adoption of electronic health record systems among providers.³¹ With the introduction of Meaningful Use guidelines for Electronic Health Records in 2010 by the Centers for Medicare and Medicaid Services (“CMS”), the conducting of a security risk analysis was added in the list of compliance measures for which health systems could obtain incentives.³²

While the laws and regulations helped with clinical care processes, they made the connection of data from clinical care to research more difficult. A report from the Institute of Medicine Committee in 2009 concluded that the HIPAA Privacy Rule does not protect privacy as well as it should and impedes important health research.³³ Also, the lack of an overall coordination between federal level privacy laws and regulations

²⁶ NAT’L INSTITUTES HEALTH, *The Precision Medicine Initiative Cohort Program – Building a Research Foundation for 21st Century Medicine* (Sept. 17, 2015), <https://www.nih.gov/sites/default/files/research-training/initiatives/pmi/pmi-working-group-report-20150917-2.pdf>.

²⁷ *Id.*

²⁸ Rachel E. Sherman & Robert M. Califf, *What We Mean When We Talk About EvGen Part II: Building Out a Nation System for Evidence Generation*, U.S. FOOD & DRUG ADMIN. (May 3, 2016), <https://blogs.fda.gov/fdavoices/index.php/2016/05/what-we-mean-when-we-talk-about-evgen-part-ii-building-out-a-national-system-for-evidence-generation/>.

²⁹ Daniel Solove, *HIPAA Turns 10: Analyzing the Past, Present and Future Impact*, 84(4) J. AHIMA 22, 22-28 (2013).

³⁰ *Id.*

³¹ *Id.*

³² *Id.*

³³ INST. MED., *Beyond the HIPAA Privacy Rule: Enhancing Privacy, Improving Health Through Research* (2009).

along with varying state laws and regulations creates a complex and sometimes contradictory set of rules to follow when conducting population health studies in many states.

III. TENSIONS

The factors accelerating population health research and those resisting change create a tension that requires resolution.³⁴ Each person has a “data privacy meter” that goes from not minding who has access to their personal health data to wanting tight control over their data at all times. Where someone sits on the meter can vary by social climate, personal risk tolerance, and by the type of data being shared. While there is significant support for open data for solving health issues, there is a countering force of ensuring a high level of privacy controls. This leads to the untenable situation where perfect privacy is only obtained when no data is shared or where open access for health research for societal benefit leads to complete loss of personal privacy. The ideal solution set will require innovation that lowers the risk or perceived risk for individuals while enabling the efficiency and quality of population health research.

The issue raised in population health research is a subset of the broader tension between the benefits and protections for the individual and the benefits and protections for society where each group is interlinked. Individuals influence a society and society influences individuals. A mutually satisfactory balance for individuals and society is an iterative process that is usually achieved by policy and technology solutions. Some of the solutions may be found within the domain of population health research and some of the solutions may come from other domains with similar individual-society tensions. Most likely, the solution framework will need to address key principles of inclusive governance and power sharing among key stakeholders and transparency regarding the process of decision making and the information and processes used to make decisions. Philosophies of social justice and human rights will need to be considered.

IV. POTENTIAL SOLUTIONS

Evolutionary Steps

Evolutionary steps involve making subtle, iterative advances from existing frameworks to finding the right balance between data sharing and data privacy. Inter-institutional trust models among entities holding and generating data and enabling that data to go through an intermediary to an end-user for research can be managed through contractual agreements. These include business associate agreements, data services agreements, data use agreements for limited data sets, and consent for sharing protected health information. However, the need to execute one off agreements for each scenario can be cumbersome. Others have begun

³⁴ Elizabeth Alexandra Gray & Jane Hyatt Thorpe, *Comparative effectiveness research and big data: balancing potential with legal and ethical considerations*, 4(1) J. COMP. EFF. RES. 61, 61-74 (2015).

creating data repositories with IRB approval and participant consent to hold data for current and future research opportunities as an alternative approach.

Disruptive Innovation Steps

Disruptive Innovation is a theory that has taken systems that are large, complicated and expensive, and through various processes, simplified, increased efficiencies and cost effectiveness of this theory.³⁵ Some potential “disruptive innovations” to improve healthcare are listed below:

Options Other than Written Informed Consent Process. More disruptive solutions have included an “opt out” vs. “opt in” research framework where persons who do not acknowledge that they do not want their health data shared will be volunteering to have their data shared.³⁶ Broad notification rather individual notification could be used along with community consultation.³⁷

Use of Privacy Standards Other than HIPAA. Another option is to use a privacy standard other than HIPAA. For instance, a framework that obligates persons and entities who collect, access and disclose information to adopt responsible data stewardship practices, regardless of whether or not data subjects have provided consent and where privacy rests primarily on the data holders has been used in the telecommunications industry.³⁸ The Fair Information Practice Principles (“FIPPS”) includes providing a framework of policies regarding openness and transparency; purpose specification; collection limitation and data minimization; use limitations; individual participation and control; data quality and integrity; security safeguards and controls; accountability and oversight; and remedies.³⁹

Compensation Programs. Another option is to reduce the risk of harm for all involved through insurance mechanisms. For instance, after lawsuits against vaccine companies and health care providers threatened to reduce vaccine supply and vaccination rates, the National Vaccine Injury Compensation Program was created in the 1980s.⁴⁰ It had a framework that rare but anticipated serious adverse events could occur with vaccines. It established a no-fault alternative to the traditional legal system for resolving vaccine injury petitions.⁴¹ A

³⁵ Jason Hwang & Clayton M. Christensen, *Disruptive Innovation In Health Care Delivery: A Framework for Business-Model Innovation*, 27(5) HEALTH AFF. 1329, 1329-35(2008).

³⁶ Deven McGraw et al., *Privacy and confidentiality in pragmatic clinical trials*, 12(5) CLINICAL TRIALS 520-29 (2015).

³⁷ *Id.*

³⁸ *Id.*

³⁹ *Id.*

⁴⁰ HEALTH RESOURCES & SERVS. ADMIN., National Vaccine Injury Compensation Program, <https://www.hrsa.gov/vaccine-compensation/index.html> (2018).

⁴¹ *Id.*

similar federal insurance structure could be created with health systems engaged in research with protected health information where even with the best controls breaches can occur. The health systems can pay into such a system and compensate individuals with the purchase of identity monitoring over time.

An alternative to paying claims after an event has occurred could be for health systems to offer identity protection insurance for any person seeking care institutions and enabling persons to access identity preservation services if a breach occurs through these commercial entities.

V. CONCLUSION

In 1979, The Belmont Report laid out the Ethical Principles for Human Research.⁴² These included respect for persons, beneficence and justice.⁴³ This framework still holds as we think about the acceleration of population health research to create a social good for all. Respect for persons includes understanding their personal privacy setting and to come to some shared decisions about when to share and not share information. Beneficence focuses of minimizing risk of harm in situations where all the outcomes are not known. Justice involves not having a single group take the risk for all that would benefit. In the end, there is significant promise in improving the value of health and in reducing suffering with the advanced technologies that generate information about the mechanisms leading to health. We still have to find the right way to advance the “commons” of population health research in order to provide health equity while maintaining the highest level of respect and responsibility for persons who volunteer to be part of population health research.

⁴² U.S. DEP'T HEALTH & HUM. SERVICES, *Ethical Principles and Guidelines for the Protection of Human Subjects of Research: The National Commission for the Protection of Human Subjects of Biomedical and Behavioral Research* (Apr. 18, 1979), <https://www.hhs.gov/ohrp/regulations-and-policy/belmont-report/index.html>.

⁴³ *Id.*