Applied & Translational Genomics 3 (2014) 79–81

Contents lists available at ScienceDirect

Applied & Translational Genomics

journal homepage: www.elsevier.com/locate/atg

Editorial

Global genomic knowledge sharing — A call for affirmative action

Introduction

In vain have you acquired knowledge, if you have not imparted it to others.
Deuteronomy Rabbah c. 900

That big genomic datasets are necessary to power translational studies would seem tautological save for the fact that sharing can be the exception not the rule. Despite market incentives that encourage data secrecy, there is growing agreement that broad data sharing is a must if molecular medicine is to exist. Further, the expanding number of global data repositories reflects the fact that disease risks are not only inborn but involve widespread life style choices or exposures, thus reinforcing our global interdependence. Thus, the promise of improving health and prevention of disease will not be fulfilled, particularly on a global scale, unless data sharing is the norm. An increasing number of initiatives are putting this sentiment into practice, some of which are presented here.

If health care is a right, and medicine necessary, then public health, global health, is a sociological and ethical as well as health problem. The US Affordable Health Care Act supports this. Arguably, it represents public affirmation of a paradigm shift, away from actuarial fairness, which bases one's obligations only on one's needs to a moral commitment to the common good. A commitment to creating a healthier, and more productive society, entails ensuring that the best available clinical knowledge is accessible to all.

The notion of benefit sharing was developed as a strategy to achieve these goals and the framework has influenced new knowledge distribution strategies (Winkler et al., 2014; James et al., 2014).

The 2002 Budapest Open Access Initiative (http://www.budapestopenaccessinitiative.org/) exemplifies a step in this direction. It established a moral imperative to advance knowledge through collaboration and share it equally among better and worse resourced entities. As such, the initiative represents a paradigm shift to a notion of distributive justice based on the reality of interdependence and a principal of solidarity. In doing so, it defined the moral underpinnings of scientific information sharing. However, the for-profit nature of the global health care industry incentivizes proprietary knowledge creation and thus legitimized the secrecy of vast amounts of genomic and related data, which remain siloed and locked to outsiders. Within these opposite but parallel strategies – open and closed data systems – lies translational research.

This special issue calls attention to conflicting norms governing data/knowledge sharing for the purpose of generating global dialogue about how best to ensure that less resourced countries can contribute to important translational research and ultimately genomic benefit sharing. It discusses current data/knowledge sharing practices, unmet needs in advancing translational genomics and collectively raises the question of what moral principles should guide data/knowledge sharing policy decisions given that the health care industry is a free market enterprise. We begin with Scully and Khoury's 'What is Translational Genomics? Going Beyond the Bedside to Improving Individual and Population Health'. The authors demonstrate why translational genomic research needs to go beyond the traditional "bench to bedside" model to include improvements in healthcare and disease prevention. They discuss four over arching phases of translational genomic research subsequent to initial discovery and demonstrate why research is needed to determine whether an innovative application is indicated for widespread routine clinical use. Further, different types of research are needed; end-user behavioral research, comparative effectiveness research, and adoption research to assess whether dissemination and appropriate clinical integration have been achieved and whether or not disparities in access have occurred. The promise of genomic medicine, they say, will not be fulfilled until these phases of translational research are conducted.

The need to close scientific knowledge gap between better resourced and less resourced areas of the world was recognized around the time that the Budapest Open Access Initiative arose. In 2001, representatives from the World Health Organization met with six of the largest international scientific publishers and established HINARI Research in Health to bridge this gap. In 'HINARI: Opening Access in Biomedicine and Health', Robertson discusses HINARI, its overall achievements and impact to date. The program is funded through 2020.

The growing demand for transparency and openness coupled with the increasing cost of drug development has prompted the pharmaceutical/biotech industry to test out a new business model. Au's article, 'The Shift to an 'Open' Model of Drug Development' reviews the major factors driving pre-competitive collaborations. Although the parameters of the pre-competitive space are not yet well defined, the value of the approach is that all participants gain the same information from a single source. For industry, this translates as faster drug development and robust pipeline.

Collaborations between research scientists and citizens extend beyond the confines of clinical research. Citizen science embodies the ethic of openness, making the process of conducting scientific research available to the 'masses' for collaboration. Dr. Curtis's article, Online Citizen Science Games: Opportunities for the Biological Sciences takes us into the world of three successful biological citizens – or crowdsourcing science – gaming formatted research projects; Foldit, Phylo and EteRNA. Each harnesses the collective problem-solving abilities of non-experts to accelerate scientific progress. Her research found that a relatively smaller number of users are active players/problem-solvers, likely due to the lengthy tutorials required and difficulty of the games. Nonetheless, participants gain a high degree of

http://dx.doi.org/10.1016/j.atg.2014.09.014

scientific knowledge and some research problems effectively solved through crowdsourcing, with scientists often learning different approaches to problem solving. Thus, these types of collaboration have been shown to be mutually beneficial while instantiating a commitment to society's common good.

Though the Open Source science movement is comparatively well developed in the US and UK, little is known about open knowledge projects in other parts of the world. Corbi and Thierry recently undertook to close this gap by traveling the globe to identify and document open knowledge projects in disparate parts of the world. 'Open Steps: a Journey to Discover and Document Open Knowledge Projects Around the Globe' describes a range of different types of open knowledge projects, their challenges and community impact. It offers a glimpse into how people in less resourced areas of the world are using newly opened databases to solve important local problems. Among the projects discussed is DNAdigest, an initiative that provides a mechanism for genomic data sharing among researchers situated in disparate research settings as well as areas of the world. DNAdigest's mission is to identify and remedy barriers to efficient and ethical genomic data sharing in the human genomic research.

In 'The need to redefine genomic data sharing: a focus on data accessibility' Shaik et al., present DNAdigest's qualitative study of clinical, academic and industry researchers' experience with accessing and sharing genomic data. Results reveal sharing barriers in discoverability and accessibility and areas of researcher's frustrations. Solution strategies are suggested.

One such solution strategy is discussed in the companion papers, 'A Collaborative Approach to Develop Multi-Omics Data Analytics Platform for Translational Research', which is the first of two papers, the second will appear in the next issue. Addressing the well known pharmaceutical R & D need to analyze integrated datasets, such as clinical patient data with high dimensional omics data, the authors describe a collaboration between GeneData's Analytic platform and the tranSMART knowledge management platform, which enables sharing, integration, standardization and analysis of heterogeneous data. The paper demonstrates the feasibility of linking academic, non-profit and corporate research communities for collaborations facilitated by tranSMART's open-source platform. Schmacher et al., describe how the integration of these platforms achieves data integration and facilitates big data analytics, thus solving a formidable obstacle to translational research.

While a growing number of countries around the world are conducting large-scale human genome sequencing and benefiting from technological innovations that solve data sharing bottlenecks, advance analysis and interpretation, overall goals will not be reached, if a flexible balance between protecting donor privacy rights and permitting data access is not established. In Data Acquisition and Data/Knowledge Sharing in Global Genomic Studies, Rotimi and Mulder discuss the importance of an approach that can overcome the challenges in less resourced areas of the globe, lest valuable data be available for use, benefit only the developed world. They boldly call for measures to improve the capacity (skills and infrastructure) of researchers in the developing world to enable their ability contribute equally. Failure to improve the skills and infrastructure for such researchers, they contend, could easily result in their data becoming publicly accessible too soon, due to funding requirements regarding data sharing, thus barring their inability to publish first, particularly in light of difficulties in processing large volumes of data because of poor internet connectivity. They call for publication embargoes that recognize these limitations. Furthermore, data sharing and access policies for primary and secondary use of data, they argue, must be fair in taking into account constraints in the developing world and involve clear and ethically robust consent process.

In 'Genomic Knowledge Sharing' Francis provides an updated review of legal and ethical issues related to genomic information sharing. Achieving the common good, she claims, is served not only by genetic information donor duties but genetic information recipient duties as well. Individuals have a moral duty to share their genomic information for the common good, provided that they know how their information is to be shared and that they are protected against risks of disclosure. Holders of individual data, she claims, also have important duties which tend to be less recognized; namely to protect and ethically use the individual data they are entrusted with. Reciprocal duties, she claims, serve both justice and the common good.

Whether regulation is needed to ensure data sharing, as well as ethical data sharing conduct is taken up by Chalmers et al., in 'To Share or Not to Share: Is the Question'. Their article examines whether an international code of data sharing conduct can change attitudes and practices towards more responsible and secure sharing of research and clinical data. The question is discussed against the promulgation of international principles governing ethical and legal conduct of genomic research over the past twenty years. Legal and ethical pitfalls in international genomic/clinical data sharing have begun to be addressed by the Global Alliance for Genomic Health (GA4GH) which is developing a Code of Conduct for International Data Sharing. Chalmers et al. argue for institutional endorsement of the ethical principles laid out in this GA4GH document. However, Chalmers et al., appropriately argue that a rush to regulate should be tempered given the absence of data on specific legal barriers.

Interviews with Drs. Gholson Lyon and Jeantine Lunshof shed light on the impact of data access data sharing practices on research progress. Their respective views drive home just how powerful the impact of existing requirements is on not only research but also, importantly, personal careers.

While many genomic databases are public and in theory freely accessible to anybody who meets access requirements, there is a growth of private companies sequencing organisms of interest. These databases, or aspects of them, may be available for a cost which academicians are normally unable to pay. Thus, a substantial amount of sequence information is available only to big companies with deep pockets. Principles of just and ethical data sharing run up against free market norms, raising the question of whether the goals of translational research and promise of precision medicine can be realized by all, globally. Angrist and Cook-Deegan address this problem in 'Distributing the Future: The Weak Justifications for Keeping Human Genomic Databases Secret and the Challenges and Opportunities in Reverse Engineering Them'. Drawing on the negative market impact of Myriad's litigation and data secrecy strategies, they discuss non-commercial efforts to recreate the secret data, and why proprietary databases, or at least parts of them, will eventually, become open. Their vision of a redistributed future in which data donation, sharing, and access not only support goals the common good but also lead to the widespread benefit sharing calls for not only a new social contract but also a reengineered genetic testing and therapeutic market. De-incentivizing data hoarding can surely benefit research and development in the less resourced parts of the world.

Regardless of how spectacularly genomics advances, if providers are not competent to use new genomic knowledge and tools in daily clinical decision-making then the field will have failed on its primary mission, to improve detection, treatment and prevention through the practice of molecular medicine. De Abrew, Dissanayake and Korf review major challenges in educating providers across the globe. Scarcity in low and middle-income countries hampers not only the affordability of technologies, but also slow adoption limits the ability to study whether technologies improve health outcomes. Lacking convincing evidence of clinical benefit, they argue, compromises education efforts. They discuss the pressing need to remedy cultural inequalities to develop minimum standards of genetic competency within the medical community. Echoing sentiments expressed by other authors of articles in this issue, they discuss why infrastructure must be built and appropriately used if less resourced clinical researcher can hope to conduct quality translational research.

Electronic health records are one element of the needed infrastructure. As Fein points out in 'Innovate of Die: Genomic data and Electronic

Health Records', innovative solutions are needed to ensure that genomic data resides within the EHR and thus serve clinical care. Ideally, the developing world will adopt solutions that optimize the integration of disparate data and avoid the mistakes and challenges that are all to well known in the US.

Finally, Dr. Al-Mulla's commentary, 'The Locked Genomes: A perspective from Arabia', presents a range of data sharing challenges faced in Kuwait. Their story is potent for it reveals both a passion to advance genome knowledge and help patients and the grim reality that without the ability to compare sequence variants, less resourced countries will remain disadvantaged in an ability to participate in global genomic research, as well as reap the benefits that are hoped to be shared globally.

The reasons to share data are increasingly evident and compelling. Novel technological solutions to permit sharing across disparate domains are increasingly available and principles governing appropriate sharing will soon lead to practice standards. We ought not be fooled, however, into believing that therefore open access, data sharing and global benefit sharing are a fait accompli. The ethos of open data/knowledge sharing exists within a free market economy that prioritizes considerations of financial gain over justice for the less resourced. A commitment to open access, global genomic data and knowledge sharing reflects a call for a new social contract based on a principle of solidarity and a duty to act for the common good. Such a new social contract would likely entail asking companies to do things contrary to their best financial interests. Therefore, to ensure a paradigm shift, strategies to financially incentivize sharing are likely required. Equally important are strategies to drive principles of beneficence and distributive justice for the less resourced. The time for principled global discussions that ensure global benefit sharing is now. Pardon the pun, but a call for affirmative action just may be the way to ensure fair play.

> "Equality of opportunity is not enough. Unless we create an environment where everyone is guaranteed some minimum capabilities through some guarantee of minimum income, education, and healthcare, we cannot say that we have fair competition. When some people have to run a 100 metre race with sandbags on their legs, the fact that no one is allowed to have a head start does not make the race fair. Equality of opportunity is absolutely necessary but not sufficient in building a genuinely fair and efficient society."
>
> [—Ha-Joon Chang, *23 Things They Don't Tell You About Capitalism*]

References

James, R., et al., 2014. Exploring pathways to trust: a tribal perspective on data sharing. Genet. Med. (May 2014) http://dx.doi.org/10.1038/gim.2014.47 (Epub ahead of print).

Winkler, S.J., et al., 2014. A distributed model: redefining robust research subject advocacy program at the Harvard clinical and translational science center. Clin. Transl. Sci. 7 (4), 329–335 (Aug 2014).

Carol Isaacson Barash Ph.D.
Principal, Helix Health Advisors Editor-in-Chief, Journal of Applied & Translational Genomics, Elsevier United States.
Corresponding author.
E-mail address: cibarash@helixhealthadvisors.com