Accepted for Handbook of Genomics, Health & Society in May 2017

Section Introduction: Genomics and DNA-Based Technologies in the Clinic and Beyond

Sabina Leonelli
University of Exeter
s.leonelli@exeter.ac.uk

The role of genomics in society has become ever more entrenched within the last decade. This is partly due to advances in technologies and particularly sequencing tools, which have transformed the act of obtaining an individual's genetic pedigree from an esoteric, labour-intensive, costly exercise to a largely automated, relatively affordable and mundane practice. It is also a result of the increasing globalisation of DNA-based technologies, which have been picked up by health systems, governments, insurance companies and data analysts all over the world, thus becoming more and more of a platform for international dialogue around understandings of health and disease. Given these developments, it is tempting to think of this historical moment as a "postgenomic" one in the sense that DNA-based tools - and related ideas about human nature and wellbeing - have irrevocably and fully established themselves across cultures and social customs, thus creating a uniform reference point for biomedical practice. And yet, talk of postgenomics can be interpreted very differently. It can indicate the increasing awareness by the public at large (including biomedical researchers and physicians) of how difficult and complex it is to interpret genetic results, and thus to create any generalized understanding of biological process and therapeutic intervention (Richardson and Stevens 2015) or ways to exploit advances in genomic understanding to effectively target the unique characteristics of individuals and groups (Green and Voigt 2016). Postgenomics can also be taken to mark the increasingly hegemonic institutional, corporate and regulatory landscapes in which DNA-based technologies are taking root (Peterson 2014, Sunder Rajan 2017, Murphy 2017), and the myriad questions surrounding their prominent status within and beyond biomedical and clinical environments around the globe (Bliss 2017, Reardon 2017). In short, postgenomics can and arguably should be viewed as a marking the tension between the growing entrenchment of genomic practices within medical practices and markets, and the enormous logistical, scientific and moral challenges posed by enacting genomic knowledge and tools across very different medical regimes, skillsets and patients.

It is becoming ever more apparent is that while technology may help enormously in making genomic sequencing and related tests increasingly affordable and available, the challenges involved in managing and interpreting the resulting data have in no way lessened. If anything, the hard problems confronted when attempting to decide who should have access to genetic data, and to do what, have acquired a new degree of severity due to the overarching shift in discourse, practices and commercial interests around big and open data (Ebeling 2016, Leonelli 2016). Calls to view, handle and value data – and particularly personal health-related data - as sources of economic and political power are proving increasingly convincing, in the face of systems of data gathering and dissemination that exclude large parts of the population, are subject to very few restrictions concerning potential misuse and misinterpretation of the data at hand, and function as systems of citizen surveillance, protection and service provision all at the same time. Furthermore, some types of data are systematically favoured over others for technical and

commercial reasons, again calling for an assessment of what such selective gaze implies for different social groups and activities. Genomic data in particular are often prioritized, thanks to their digital, portable format, their embedding in well-entrenched and highly marketable medical technologies and ways of knowing, and their continuing privileged status as quantitative documents of people's biological inheritance.

What at the turn of the century was mostly a worry for technical experts is now becoming acknowledged in public discourse: creating data may be relatively easy, but preserving them, protecting them and analysing them is difficult and expensive. Even more difficult is deciding who has the expertise and power to take care of such data and use them to extract biomedical insights, ground political and economic decisions, and shape perspectives on the future – and what responsibilities and accountabilities are involved in this process. This is particularly true in a world where internet communication and related technologies are ubiquitous, and yet the capacities to exploit those technologies are very unevenly distributed, and the ways in which they manifest themselves in specific situations, across geographical locations and cultural norms, continues to vary enormously. Contemporary information and computing technologies may make it easier for people to communicate, but who is communicating what to whom around the use of genetics in society, and with which results, interpretations and purposes? And how does such communication unfold in the contemporary political context, where nationalism and populism are on the rise in several high-income countries, and attitudes to national borders, cultural diversity, the threat posed by movements of people and related biological materials, and the legitimacy of scientific expertise are shifting?

These are the question that motivate the first section of this new handbook, where authors examine the most recent developments in genomic and DNA-based health technologies, and bring fresh perspective on their social and scientific role that takes account of the evolving political landscape. Catherine Bliss starts off the section with a comprehensive review of contemporary instantiations of *biomedicalization*, which she proposes as the lens that "helps us ascertain the major shifts in today's social order around the expansion of biomedicine." Her discussion includes the role and goals of medical care and the pervasiveness of molecular conceptions of biomedicine in everyday life, with significant implications for conceptualisations of the body, personal identity and the pathological. Her chapter situates the development of DNA-based technologies within a rich social, cultural, economic and political context, thus providing a textured landscape for many of the themes that other Handbook contributors will address in some detail in what follows.

The chapter by Niccolo Tempini and Sabina Leonelli then zooms into the ways in which the emergence of big data discourse, infrastructures and practices has affected – and, arguably, boosted - the role of genomics in biomedical research and care. They emphasise the ways in which attention to data practices shifts the focus of STS researchers interested in biomedicine beyond the clinic, to embrace the vast variety and multiplicity of social environments (digital or physical) in which the production, dissemination and interpretation of data of medical relevance is happening, and in which data practices are valued in a variety of different ways by different actors. They conclude that whether and how the transformative promise of big data can be delivered for biomedicine and healthcare depends on the tools and assumptions used when assembling, interlinking and integrating genomic data with other types of biomedical data – a task fraught with technical, ethical and social challenges.

Susan Kelly, Anna Harris and Sally Wyatt also place emphasis on the ways in which genomics is escaping the biomedical and clinical context, by examining in detail the rise and commercialisation of personal genetic testing. Drawing from studies of the dynamics and usage

of the internet, as well as research on the enactment of direct-to-consumer testing by online providers such as 23andMe, this chapter stresses the extent to which bringing genetics out of the clinic is impacting the identity of patients (who are at once consumers and contributors of a service with multiple goals and accountabilities), their relationship with healthcare professionals and their understanding of the value of data produced through medical interactions. In particular, Kelly, Harris and Wyatt emphasise the disruptive effect of this evolving landscape on the various relations of trust that underpin and facilitate biomedical knowledge and care.

What do these developments mean for clinical work? This question is tackled by Ilana Lowy's chapter, which builds on the history of genetics and its clinical applications to portray how genetic testing and counselling practices have evolved over the last fifty years, the relation between such developments and broader societal trends, and the effects of these shifts on contemporary clinical practice. Lowy focuses on the increasing diversity of tests and targets developed for clinical use, the strong link between test availability and patients' uptake, and the implications for understandings of risk, parenthood and parental responsibility. She also points to the challenges raised by unanticipated secondary findings, as for instance in the case of hereditary conditions that affect whole families, and the resulting climate of "managed fear" in which human reproduction is now planned and enacted.

The last chapter in the section moves away from the Global North and Anglo-American trends, and takes a close look at the historical development and contemporary social implications of the implementation of genomic technologies in China. Jianfeng Zhu, Shiyi Xiong and Dong Dong focus specifically on prenatal genetic testing, a particularly sensitive issue in China given its birth control policies as well as the Confucian approach to responsibility and bonds within the family. The chapter considers recent changes in governmental discourse around the regulation of maternal and infant care, and particularly the role played by genetic knowledge in the current shift from a policy centred on population "quality control" to an opening towards personal choice around "reproductive insurance". As the authors point out, this intersects in complex ways with the expectations and preferences of the families affected by state policies, as well as the training and everyday practices of healthcare professionals tasked with delivering treatment and assistance. It also exemplifies the surprising speed with which a country can reposition its role as participant and contributor to global health discourse and practices, with China now rapidly moving to establish genomic collection facilities to capture data and samples from its population and make them visible and potentially accessible internationally.

Genomic practices continue to be caught in a web of technological acceleration, societal changes and logistical chaos, with financial resources and market forces driving both the direction and the location of innovation in medical care. As pointed out in all the chapters within this section, this has substantial and uneven effects on popular attitudes and discourse on reproductive technologies and genetic testing. In particular, the increasing alignment of service provision, commercialisation and medical care derives in confusion around who is responsible and trustworthy, for what and in which way – a confusion that affects not only prospective patients, but also healthcare professionals and regulators. In this moment of transition and change, it is critical for scholarship in the history, philosophy and social studies of genomics and biomedicine to support the development of civil epistemologies around biomedicalization, and inform emerging regulatory efforts, legal frameworks and commercial strategies.

Acknowledgments

Funding from the European Research Council (grant award 335925) and the UK Economic and Social Research Council (award ES/P011489/1) has supported the research on which this introduction is grounded. I am grateful to the Handbook's editors, especially Sahra Gibbon, Janelle Lamoreaux and Barbara Prainsack, for their excellent support, prompt assistance and helpful suggestions throughout; the authors of the chapters in the section for their willingness to work to a tight schedule; and to Aaron Parkhurst for precious editorial assistance.

References

Bliss, C. 2017. *Social by Nature: How Sociogenomics is Redefining What it Means to be Human*. Stanford University Press.

Green, S. and Voigt, H. 2016. Personalising Medicine: Disease Prevention in Silico and in Socio. *Humana.mente Journal of Philosophical Studies* 30: 105-145.

Ebeling, M. 2016. *Healthcare and Big Data: Digital Spectres and Phanthom Objects*. Palgrave McMillan.

Leonelli, S. 2016. *Data-Centric Biology: A Philosophical Study*. Chicago, IL: Chicago University Press.

Murphy, M. 2017. *The Economization of Life*. Duke University Press.

Reardon, J. 2017. *The Postgenomic Condition: Ethics, Justice, Knowledge After the Genome*. Chicago, IL: Chicago University Press.

Richardson, S. and Stevens, H. 2015. *Postgenomics*. Duke University Press.

Sunder Rajan, K. 2017. *Pharmocracy*. Duke University Press.