Methods Futures Briefing #001

Human Social Genomics

By Robert Meckin[1] and Mark Elliot

This Methods Futures Briefing focuses on aspects of human social genomics that have a bearing on social research claims and methods. It first outlines definition of genomics and some key findings, promises, developments when those methods are used in the context of human populations. The following section discusses potential social research methods-related issues that arise from development and deployment of human social genomics and closes with a brief consideration of the future of genomics and social research.

What is human social genomics?

The entire genetic sequence information of a particular organism is known as a genome and genomics focuses on genome-level data comparing variations in gene regulation and expression. **Genomics** broadly studies the interactions between genome, environment, and genetic outcomes for an organism in question, and so represents a shift in understanding from deterministic individual genes to reactive, unstable genomes (Fox Keller, 2014; Slavich and Cole, 2013). Genomics can also include **epigenomics** which is the study of the chemical 'tags' that mark an organism's genome, changing the ways that genes are expressed in response to contextual exposures and life experiences of the organism.

Human social genomics studies the relationship between gene expression and social conditions of humans (Cole, 2014). Socio-environmental factors have been shown to affect expression of ten times as many genes as genetic factors in leukocytes genomes (Idaghdour et al., 2010) indicating the importance of environmental context for human genome function. Terms like human social genomics and **social science genomics** are terms used to refer to the combination of social and genomic knowledge and research in humans e.g. (Conley and Fletcher, 2017; Freese, 2018).

Human social genomics may be particularly useful in understanding how 'social experiences, like those associated with socioeconomic status (SES), affect physical and mental health' through effects on molecular activity in cells (Shanahan, 2013: 256). Such approaches have been heralded as having great potential for human health (Slavich et al., 2023).

Methods

Genome-wide association studies (GWAS) are a primary methodology within genomics and find associations between gene variants and diseases. GWAS test different gene variants (polymorphisms) in different genomes. **Genome wide transcription profiles** show genes expressed in individuals (Cole et al., 2007). **Multilevel analyses** help link polymorphisms to concepts such as health, SES, and life course that may be of interest to demographers and social epidemiologists (Slavich et al., 2023; Shanahan, 2013). Population-based **genome sequencing studies**, which are aimed discovering rarer polymorphisms (often related to disease) are becoming more common (Hindorff et al., 2018).

Policy and data in the UK

The UK has a flagship policy 'Genome UK' focused on using genomics for health (HM Government, 2020). The UK also has a history of considering genomics, social science, and society (Government Office for Science, 2022; Diamond and Woodgate, 2005). Since 2006, the UK biobank has been collecting genomic, biological, demographic and personal data from 500,000 citizens aged 40-69 since 2006. Researchers can apply for access to this anonymised dataset to explore different questions arising from the interaction between biology and environment. It has been referred to as 'the world's most important health database and arguably the UK's most significant scientific asset' (Sample 2023). Another

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large multi-sector programme was, Our Future Health, began recruiting the first tranche of its planned 5M-strong participant cohort in 2022.

Critical issues for social genomics with social research

Social genomics goes right to the heart of methodological philosophy. The 'big question of nature vs. nurture has taken on a new significance' (Goisauf et al., 2020: 2). This is because debates about **biosocial research** centre on ontology of the social world and what we can know about those categorisations. Such debates also deal with determinism, causality and ethics (Meloni, 2015). They have led to deep rifts in the field of anthropology and between academics within specific institutions (Jumonville, 2002; Keller, 2016).

An example here is what happens when socially constructed categories of race, ethnicity, or sexuality are treated as absolute factors in statistical analyses (Morning, 2014; Meloni et al., 2022; Goisauf et al., 2020). Genomics methods that objectify the genome become problematic when used to incorporate socially constructed categories such as sexuality. Furthermore, 'claims to working under the premise of 'pure science' of genomics are untenable as the social is present by default—within the methodological choices made by the researchers, the impact on/of the social imaginary or epigenetic context' (Goisauf et al., 2020: 7). In other words, social genomic claims should demonstrate an awareness their own practices, assumptions and ethics, and show sensitivity to context and the complexity of social life – perhaps through integrated or facilitated **reflexivity** – in order to be robust across disciplines.

There is a related methodological tension. To avoid statistical problems associated with variation, genome studies frequently lack diversity, reproducing the under-representation of particular groups, with **genomic research focuses overwhelmingly on dominant European groups** (Hindorff et al., 2018; Rebbeck et al., 2022). Total GWAS participant diversity can tracked at the GWAS diversity monitor (Mills and Rahal, 2020). For example, 88% of GWAS data are from cohorts of European ancestry with 72% of participants are from the USA, UK and Iceland meaning that portability of findings across other populations is limited (Mills and Rahal, 2019; Mills and Rahal, 2020). Thus, finding **participatory and inclusive** ways to involve publics, groups and communities in research policy and research projects, especially for under-represented groups and rare diseases, is paramount (Middleton et al., 2014; Mills and Rahal, 2019; Rebbeck et al., 2022; Young et al., 2022). At the same time, improving representation, participation, and diversity needs to happen without committing to a methodological reification of particular social categorisations.

Given the size of the databases and information, genomics is often cited as an example of **big data** with implications for how knowledge is imagined to made because it is often associated with ideas of 'pure data' and 'more objective' data (Goisauf et al., 2020). Findings can be framed causally when they correlations are being described (see *Critical questions for big data* (boyd and Crawford, 2012)).

Human social genomics invokes **interdisciplinary collaborations**, which raises the issues of how researchers work together and how methods are chosen, undertaken, and valued. Researchers may use the language of translation to justify social science of and with genomics (e.g. (Burke et al., 2015; McBride et al., 2010), which might suggest that genomics follows an independent trajectory. This suggests the social sciences take a service role (Barry and Born, 2013). Collaborating disciplines may need to methodologically recognise different power relations that affect knowledge production.

A future with increasing social genomics

Advances in genomic data generation, storage, and processing, mean that genomic datasets may grow and become more complex. Coupled with policy emphases on scientific leadership in biomedical health (e.g., UK biobank) and attention to global health challenges, human social genomics and cognate to approaches to health **may well expand**.

Continued **efforts to monitor and improve participation, diversity and inclusion** are likely, with the aims of making human social genomics findings more representative of, and transportable between, particular groups and populations.

Training and **awareness building of histories of biosocial debates** and of the ethical implications of ontological and epistemological choices are required so that potential collaborators are aware of the philosophical differences that are generated in genomic and some social approaches to knowledge production.

There is potential that social genomics collaborations develop new ways of taking account of the social by using methodologies that reflect the socially constructed nature of genomics knowledge, especially where GWAS seek to make claims about the social.

A further consequence of social genomics is to treat genomes at a community or population level, that is produced by genetic processes mediated by social process and potentially multiple species, rather than the level of individual organisms (Slavich and Cole, 2013). This would have clear implications for human-nonhuman relationships. A **metagenomics** of this kind, however, is in its infancy.

Additional reading

NCRM produced two interface reports focusing on computational social science (Meckin and Elliot, 2021a) and social and health research (Meckin and Elliot, 2021b), respectively, and these contain information and analyses relevant to this briefing.

If you would like to contribute a Methods Futures Briefing to the series, or would like to give feedback, please get in touch by emailing Robert.meckin@manchester.ac.uk.

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