

Genomics and Genetic Medicine: Pathways to Global Health?

This special issue of *Anthropology and Medicine* contributes to emerging anthropological research examining the expanding terrain of genomic research and genetic medicine as a product of and vector for globalisation (Beaudevin and Pordie 2016) and the ways it is being aligned with ‘Global Health’ (Koplan et al 2009). It builds on a growing body of literature in anthropology that observes how this arena of science and medicine is unfolding across a range of national and transnational contexts with uneven and often inequitable consequences (Whitmarsh 2008, Fullwiley 2011, Sleeboom-Faulkner 2010, Wade et al 2014, Taussig and Gibbon 2014). The papers presented here explore comparatively and transnationally the complex and somewhat paradoxical interface between genomics and/or genetic medicine and the emerging landscape of ‘Global Health’. It is important to note that the meeting point between these domains is not new, with the WHO consistently highlighting the relevance of genetics to addressing human health since the 1950s.¹ Nevertheless, the recent expansion of genomics as a global and globalising research terrain marks a particular moment in the way that a domain of genetic knowledge and technology is being used to address a range of health care challenges. This now encompasses not only ‘rare’ disease but also the growing rates of non-communicable chronic disease in low- and middle-income countries, as well as infectious disease, with genetics becoming tied to large transnational epidemiological studies to address epidemics such as Malaria (see Achidi et al 2008). The way that genomics and genetic medicine are being configured as a pathway to ‘Global Health’ in comparative national arenas beyond, but also often in close interaction with the ‘global north’, is of central importance for medical anthropology.

The six papers included in this edition provide a multi-dimensional comparative perspective on how a focus on genomics and genetic medicine is being aligned with public health, discourses of prevention, questions of population difference, national sovereignty and processes of nationalisation. In doing so they are attentive to how structural inequities, issues of social (in)justice, and an emerging articulation of ‘rights’ to health services and resources shape these developments. While the specificity of national context is foregrounded in articles that examine how genomics and genetic medicine is unfolding in Brazil, Japan, China, United Arab Emirates (UAE), India and Qatar, we also see how these are related, often in disjunctured ways, to transnational parameters, as this concerns funding or research priorities or international ethical guidelines for research. While standardisation of the goals of genomic research or the

¹ See Gaudillière, J.P. and Beaudevin, C.P. (2014-19) ‘Global health, From international public health to global health’ <http://globhealth.vjf.cnrs.fr> for further discussion of the history of and transformation of genomics in global health.

practices of genetic medicine is a component of these developments, it is not the only possibility or outcome (Gibbon et al. *forthcoming* 2018). The articles brought together here illustrate how a vast range of different research practices and techniques of health care intervention are increasingly encompassed under the aegis of genetics; an expansion that is happening at a time when the explanatory power of ‘the gene’ is being questioned but also reframed as an agentic component of gene-environment interactions in the emerging context of epigenetic research (Lock 2013). These practices now include research and medical interest in ‘rare’ or ‘intractable’ genetic diseases, population genetic screening and public health interventions aimed at ‘prevention’ as well as biobanking initiatives that may be targeted at specific populations. A number of the papers also attend to how medical genetics and genomics are unfolding and are being made relevant to chronic or common diseases, such as diabetes or cancer. In the social arenas outlined in the papers presented here, these are processes which are variously informed by particular cosmologies of the body and ill health, the uncertainties of rapid urban change, as well as socio-historic shifts in how legal ‘rights’ to health are being configured.

In short, the papers in this collection examine the diverse expressions, dynamics and tensions in the way that different fields of genetic medicine and genomic research are being shaped by and helping to influence processes of globalisation. They provide a rich and engaging perspective contributing to emerging and ongoing anthropological examination of genomic research and medicine in underexamined regions within the ‘global south’. In outlining the special issue in this introduction, we draw attention to three cross-cutting themes in how genetics and genomic medicine is being situated as a potential yet often still often elusive pathway in the pursuit of and at the interface with the shape shifting terrain of ‘Global Health’. This includes themes of inequalities and social justice, cosmology and population difference.

Inequalities and social justice

With calls to bridge the so-called ‘genomic health divide’ through economic investment in research and expanding the provision of genomic services and technologies to the ‘global south’, the issue of genomics and health inequalities has also become part of the landscape in which these developments are being shaped. ‘Genomics for the World’ was the headline of an article in *Nature Genetics* in 2011, where calls were made to widen the scope of genomics to include a greater diversity, to encompass ‘minority populations’ from ‘other ethnic groups’, and to ensure that ‘those most in need’ are ‘not the last to receive the benefits of genetic research’ (Bustamante et al 2011:1651, see also Popejay and Fullerton 2016). The way a discourse of ‘humanitarianism’ is being foregrounded in relation to efforts to ensure those in the ‘global south’ are given the

‘right’ to become participants in *and* potential beneficiaries of genomic research demands critical and engaged attention (Fullwiley and Gibbon *forthcoming* 2018, Reardon 2012). Yet, in this moment of global expansion and ethical repositioning of genetic medicine and genomic technologies in terms of social inclusion and justice, the reality of inequitable and stratified access and rights to health care resources is also becoming apparent. Emerging research is demonstrating how different legal or policy infrastructures that make public health possible or delimit it in certain ways also shape how ‘rights’ to health care, increasingly including medications, treatment and screening interventions for genetic disease, are being pursued (Biehl 2013). In other cases, we see how these developments are informed by colonial and post-colonial histories, particularly in how populations are defined and made targets of genetic research and/or recipients of genetic medicine (Fullwiley 2011). But histories of eugenics and their ongoing contemporary manifestations, in terms of, for instance, embryo-selection practices in certain regions of the world, also have consequences for how the complex relationship between genomics and disabilities are unfolding. This is particularly so when a discourse of prevention is being fuelled by the expanding possibilities of prenatal screening. As Scully points out, the discourses and practices of ‘othering’ at stake in the expanding terrains of genomic research and medicine are often ‘subtle’ and ‘dispersed’ (*forthcoming* 2018), demanding careful and nuanced social-science attention.

Genetic Cosmologies

The papers reveal a multiplicity of discourses, where both the global and the local participate in constituting the meaning, relevance and scope of genetics in different contexts; this means paying close attention to the way that different cosmologies are evoked, instrumentalised and transformed. Genomics and genetic medicine are often articulated to be novel and cutting-edge, but they also ironically point to tradition and inheritance. Profoundly meaningful, genetic understandings provide powerful idioms for the expression of individual and collective identity (Nash 2015; Nelson 2016). Whilst providing language for their expression, genetic discourses also contain potent possibilities for eliciting change in individual and social identity (Rabinow 1996). Increased genetic knowledge and the outcomes of its applications can impact the social fabric of society by impacting ideas of family (i.e. Panter-Brick 1991) and inheritance; they may become novel signifiers of identity, history and nationalism (Beaudevin 2013:185). The nation appears in discussions around genomic patrimony and sovereignty (Rabinow 1999; Benjamin 2009): the economics of ‘biovalue’ (Waldby 2002) mean that DNA may be regarded as a national resource, as well as a repository of national characteristics. Genetic medicine and research can be harnessed as part of national aspirations and positionings as well as part of the state’s dedication to improving the lives of its citizens; but they also engage with global linkages beyond the nation.

The contributors examine the expanding terrain of genomic research as a process,

which is characterized by both standardisation of knowledge and technologies and simultaneously locally configured. As genetic discourse develops in particular contexts, rather than replace cultural models, medical and scientific concepts are re-shaped and interpreted through local cosmologies, with tensions and negotiations often found at these junctures. As Shaw points out in her examination of British Pakistanis encountering genetic services in the UK, genetic explanations ‘provide a mechanism, but probabilistic concepts of chance leave a void that appeals to God’s will serve to fill’ (2009: 147). At the same time, and in various contexts genetic disorders are attributed to forces beyond human understanding, such as fate, destiny, and gods, as if in a kind of biological predestination (Featherstone et al. 2006: 113). Scientific notions of fate contained in genetic explanations and idioms intersect with traditional and religious conception of fate (i.e. Raz 2005) and when this occurs, both models are dynamically influenced, with novel meaning given to ideas of the body, inheritance, fate and kinship. Indeed, genetics has and continues to inform and complicate debates about what it means to ‘be in the world’ (Franklin, 1995).

The fault lines between genetic populations

New dilemmas have emerged around studies of the human genome and its parallels, the proteome and the epigenome, where the ultimate stated aim is to serve the development of global medicine as the provision of genetically specific medicine for certain populations. The ways in which genetic populations are defined, however, are closely linked to biopolitical issues, turning the definitions of genetic populations into fault lines among groups of people. Thus, battles are fought by patient groups to have their ‘genetic condition’ included into global or local research, whereby critique of race-, ancestor- and gender-based exclusion is rife (Epstein 2007; Fujimura and Rajagopalan 2011; Gibbon 2016). In other contexts, however, worries about being included as a target for genetic medicine and experimentation are expressed, with fierce arguments being launched against the racialisation of population and in some cases the cleansing of particular populations of disabled peoples (Lewontin 2001; Duster 2006; Leach Scully 2008; Kato 2010). And the science policies of some governments support genetic medicine to save healthcare spending by addressing the ‘problem’ of genetic conditions at their ‘root’. Such diverse approaches to genetic populations mark their controversial nature.

Important to the development of genetic medicine is the study of the role of **genetic** factors in determining health and disease in families and in populations, or genetic epidemiology. Genetic epidemiologists usually refer to socio-political notions, such as gender, race and ethnicity and topographic terms in defining the analytical units of their research (M’Charek 2005; Fullwiley 2008). Such definitions are at once scientific and political when making societal processes, such as migration history and kinship

systems, genetically relevant to their research, even *before* the research has even taken place (Lewontin 2001). A main aim of genetic epidemiology is to understand the relation between a population's genetic make-up and the environment. In the case of a genetic predisposition for Diabetes or Sickle Cell Disease, for instance, this can enable the state to implement policies with the aim of changing people's every-day or mating behaviour through educational and screening measures (Patra & Sleeboom-Faulkner 2010; Sleeboom-Faulkner 2011). In short, no approach to genetic medicine is politically straightforward; they are intimately linked to issues of social justice and equality (Sleeboom-Faulkner 2007; Fullwiley & Gibbon *forthcoming* 2018), cultural politics (Sleeboom-Faulkner 2006; Gibbon & Novas 2007; Kato & Sleeboom-Faulkner 2009; Kuo 2011; Kilshaw et al 2015), imagined genetic communities (Simpson 2000; Wade et al 2015) and financial gain (Smart et al 2006).

The papers

As the synopses below illustrate, the papers in this volume illuminate how genomics and genetic medicine is an arena which is shaped and itself is transformed by relations of inequality and social justice, by differing cosmologies and by how populations are defined and redefined by actors involved and implicated in these terrains of research and health care.

Sahra Gibbon and Waleska Aureliano in their *Inclusion and Exclusion in the Globalisation of Genomics; the case of rare genetic disease in Brazil* show how rare disease in the globalisation of genomic medicine is locally constituted and remade. This paper nuances debates on the politics of difference that link the coining of rare diseases with particular patient rights to healthcare facilities and investment in the life sciences (e.g., Epstein 2007). It does so by showing how in a context of severely constrained healthcare resources, and where the specificity of genetic diseases whose aetiologies and population frequencies are often contested or unknown, rareness can become a means to promote social justice and health care rights. While in the US, race and gender appear to more explicitly shape the inclusion of populations in medical research, in Brazil this process is also informed by 'judicialisation': 'a politics predicated on efforts by patients, families and patient support organisations to oblige the state to meet a constitutional commitment to provide health care to all'. Outlining how interest in rare genetic disease has emerged in dialogue with paediatrics and oncology and in relation to a partial and inequitably distributed system of public health, the authors illustrate the complex feedback loops between clinical care and research, patient mobilisation and recent state legislation. The genetic conditions of interest in their paper, centred on cancer genetic syndromes and a form of hereditary ataxia, reveal how the 'rareness' of disease has been diversely situated and differently politicized. Illustrating how clinicians, patients and their

families grapple with the boundaries between research and the right to health as well as the necessity and limits of care, therapy and prevention, the authors show how the dynamics of inclusion and exclusion play out differently, even paradoxically across and within these different arenas of genetic medicine.

Sangeeta Chatto in *Inherited blood disorders, genetic risk and global public health: framing 'birth defects' as preventable in India* explores what she describes as an 'ethical moment' in the WHO re-classification of Inherited Blood Disorders or IBD's (such as Sickle Cell Disease and Thalassaemia) as 'preventable'. This intervention is she argues part of a 'globalising turn' in the way that IBD's are being framed as a 'global health crisis' and in the way that genomics is being used to address structural inequalities that inform and shape health outcomes in low and middle-income countries (LMICs). Taking India as one social arena for these developments (see Beaudevin 2013 for an examination of these developments in Oman) she illustrates how specific colonial histories of racial classification, as this relates to caste and tribe, shape how a globalising discourse of risk relating to IBD's is mapped to certain populations and makes them 'bioavailable'. Given histories of intervention by the Indian state to control the reproductive capacity of certain ethnically marginalized communities through coercive use of abortion and sterilisation, and ongoing evidence of sex selective terminations of female fetuses, the implications of framing and intervening on IBD's in this way are profound. The challenge for public health in LMICs, as much as elsewhere is, as she points out, to engage with biological difference, disease variability and molecular heterogeneity while being attentive to the ever-present possibilities of essentialising and de-politicising health inequalities. In a context where there are dense intersections between gender, race, genetics, stratified reproduction and disability we need, she argues, to urgently attend to the 'erasures' at stake in situating IBD's as preventable.

Susie Kilshaw's article *Qatari Intersections with Global Genetics Research and Discourse* illustrates how a focus on genetic discourses is part of national and nationalising aspirations for modernity in Qatar. Genetic medicine and research is at the forefront of the state's mission to be a world leader in research, education and international politics and provide cutting-edge approaches to health care for the country. At the same time this nevertheless rests on a definition of the 'nation' that excludes significant section of the population, such as Qatar's immigrant underclass of construction workers. These 'national' aspirations also co-exist and sit in tension with a traditional emphasis on inheritance and are often in contention with the way that local Qatari's themselves understand and incorporate genetic knowledge into their lives. In this sense she, like Aaron Parkhurst (this edition), demonstrates how different cosmologies, that aspire to the modern and which reinvigorate traditional values, are characterised by a constant movement and process of re-invention as the country and its people negotiate

the new possibilities afforded by genomic knowledge and technologies. In exploring the way local Qatari systems of knowledge intersect with modernising genetic discourses the paper highlights the unevenness of change. Kilshaw outlines how ‘close’ family marriage and the traditional practice of marrying cousins, provide a particular lens for examining how cosmopolitan and traditional values and practices are aligned or negotiated and become strategies for ‘containment’ in relation to discourse of genetic risk. As others have explored elsewhere (see for instance Shaw 2015) she demonstrates how discourses and practices of consanguinity are at the heart of these intersections, illuminating how issues of modernity and tradition are being negotiated and/or contested in a variety of global contexts. In the Qatari contexts that Kilshaw investigates there are a multiplicity of discourses where both the global and the local participate in constituting the meaning of genetics and where ‘global frictions’ (Tsing 2005) are just one possibility.

Aaron Parkhurst in his *City and Cosmology: Genetics, Health, and Urban living in Dubai* discusses the role of modernity and global flows in perceptions of the health of society through his ethnography in the UAE. In this context, there is an increased susceptibility of nationals to some diseases, such as diabetes, with genetic discourses emerging in approaches to combating these conditions. However, despite widespread dissemination of genomic knowledge and its general public acceptance, the rates of diabetes, obesity and heart disease continue to increase. Parkhurst sets out to shed light on this seeming contradiction by exploring the clash between indigenous and imported biomedical knowledge systems and does so with specific reference to the city and rapid urban growth. He argues that chronic illness in the Emirates is partly the result of the ways in which many local people define what modernity means to them. Some conditions, like diabetes, are understood to have been brought by the West and represent ambivalence around modernity and outside influences. Such conditions, however, are also understood to result from a loss of valued traditional values. In the UAE, genes have been incorporated into indigenous cosmology: the language and rhetoric that his participants apply to discourses of fate are re-appropriated to help them think through genetics and other biomedical body knowledge. Instead of usurping them, a public health emphasis on the ‘Arab Genome’ has instead reinforced cosmological understandings of disease. Parkhurst suggests new local understandings of genetics give novel meanings to inheritance and share an intimate relationship with traditional ideas of fate and national identity. In the context of rapid urban growth, structures of cosmology are relied upon to cope with radical uncertainty and are applied to the body and to emergent biomedical categories. He argues that health care policy and planning should be informed by local cosmology and particularly be cognizant of how people ‘pivot themselves against a world’ that is shifting.

Masae Kato's paper *Genomics and Cure: Understanding narratives of patients with Duchenne Muscular Dystrophy in Japan* questions the presumption that all patients with a serious intractable disease are looking to genetic medicine for a cure for their condition. Having conducted in-depth interviews with patients with Duchenne Muscular Dystrophy (DMD), Kato observed how DMD patients perceive medical and scientific attempts at creating genetic medicine. She carefully outlines how attitudes towards genetics in Japan, are not only closely related to the backlash against the history of eugenics in that country (Matsubara 2003; Kato & Sleeboom-Faulkner 2017), but also to the lives and identities that individuals with DMD have built. In other words, the idea of finding a cure may not always be attractive to individuals with a condition, if this means that their life, identity and values may alter radically as a result of genetic therapy. Furthermore, whether individuals desire a genetic cure also depend on the socio-cultural conditions, such as acceptance by society, employment conditions, availability of medical and care, and the healthcare provided in a country. In brief, this case-study of DMD patients in Japan shows that patients, depending on their local circumstances and needs, will value genomic medicine differently. It serves as a warning against a widespread bias that the development of genetic medicine, rather than other medicine or facilities, are in the interest of patients with serious, intractable diseases.

Margaret Sleeboom-Faulkner's timely contribution *The justification of studies in genetic epidemiology – political scaling in China Medical City*, drawing on research conducted in Taizhou, China, argues that the way science is organized and financed leads to the strategic practice of 'political scaling' among scientists, who zoom in on particular relations between communities and research. Effectively, an intervention itself, the paper's aim is not to understand genetic epidemiology as a cultural system but rather to influence/critique its practice and conceptual assumptions. Her discussion provides a lens to understand issues and scientific practices about biobanking in China and knowledge-making in epidemiology in general. Political scaling in genetic epidemiology research means that research is justified by speculation on the relevance of data on a relatively small sample population to much wider populations, or overgeneralisation. Constituting a mismatch of scales in relevance, reach and research ethics of a study means losing sight of how the study links sample donating and benefiting communities. In the case of the Taizhou Longitudinal database, the GE study is largely supported by state institutions and justified in terms of state policies. However, it is also dependent on international scientific discourses through which it gains international recognition. The implications for the study design of this are that populations need to be defined and generalized that conform to aims of both investors and scientific audiences at home and abroad. Sleeboom-Faulkner clearly shows how anthropological fieldwork can play a complementary role to genetic epidemiology by examining the particular discourses of which a project is part.

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