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Special Issue: Genomics and Genetic Medicine

2018-05-29 10:55:13

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Anthropology & Medicine (Vol 25 [1]) brings us a special issue on Genomics and Genetic Medicine, edited by Sahra Gibbon, Susie Kilshaw & Margaret Sleeboom-Faulkner. See below for the abstracts!

Genomics and genetic medicine: pathways to global health? (*open access*)
Sahra Gibbon, Susie Kilshaw & Margaret Sleeboom-Faulkner

[Excerpt] 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 2013). 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. 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 (LMICs), 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.

Inclusion and exclusion in the globalisation of genomics: the case of rare

genetic disease in Brazil (*open access*)
Sahra Gibbon & Waleska Aureliano

Within the context of a globalising agenda for genetic research where 'global health' is increasingly seen as necessarily informed by and having to account for genomics, the focus on rare genetic diseases is becoming prominent. Drawing from ethnographic research carried out separately by both authors in Brazil, this paper examines how an emerging focus on two different arenas of rare genetic disease, cancer genetics and a class of degenerative neurological diseases known as Ataxias, is subject to and a product of the dynamics of inclusion and exclusion as this concerns participation in research and access to health care. It examines how in these different cases 'rarenesss' has been diversely situated and differently politicised and how clinicians, patients and their families grapple with the slippery boundaries between research, rights to health and the limits of care, therapy or prevention. It illustrates how attention to rare genetic disease in Brazil emerges at the intersection of a particular history of genetic research and public health infrastructure, densely complicated feedback loops between clinical care and research, patient mobilisation around the 'judicialisation' of health and recent state legislation regarding rare disease in Brazil. It highlights the relevance of local configurations in the way rare genetic disease is being made relevant for and by different communities.

Inherited blood disorders, genetic risk and global public health: framing 'birth defects' as preventable in India (*open access*)
Sangeeta Chattoo

This paper engages critically with the global assemblage framing sickle cell and thalassaemia disorders as a 'global health crisis'; and the promise of genomics, largely DNA-based carrier/pre-conceptual screening, prenatal diagnosis with a view to terminations, deployed in framing a solution to these historically racialised spectrum of diseases as essentially preventable. Sickle cell and thalassaemia are recessively inherited, potentially life-threatening haemoglobin disorders with significant variation of severity, often needing life-long treatment. I argue that the re-classification of inherited blood disorders (IBDs) under 'prevention and management of birth defects' by the WHO in 2010 can be read as an ethical moment within the 'globalising turn' of IBDs and the use of genomics in addressing structural inequalities underpinning health in low- and middle-income countries. Using an Indian case study, the paper aims at first examining the language of risk through which genes and IBDs are mapped onto pre-existing populations (e.g. caste and tribe) as discrete, categories. Second, it discusses the likely social and ethical ramifications of classifying these recessive gene disorders as essentially preventable, despite cheaply available diagnostic tests and treatment options available

in most countries in the South.

Qatari intersections with global genetics research and discourse
Susie Kilshaw

Genetic discourses have taken a predominant role in approaches to combating a number of conditions that affect Qataris. This paper is derived from an exploration of Qatari encounters with globalizing discourses of genetics, particularly as they relate to notions of risk. It explores Qataris negotiations of global interactions and influences, including the discourses around genetic risk and cousin marriage. It suggests that family marriage can be seen as one of the main platforms of resistance and a means for modern, cosmopolitan and tradition to be negotiated.

City and cosmology: genetics, health, and urban living in Dubai
Aaron Parkhurst

In light of increasingly high rates of diabetes, heart disease, and obesity among citizens of the Arabian Gulf, popular health discourse in the region has emphasised the emergent Arab genome as the primary etiological basis of major health conditions. However, after many years of public dissemination of genomic knowledge in the region, and widespread acceptance of this knowledge among Gulf Arab citizens, the rates of chronic illness continue to increase. This paper briefly explores the clash between indigenous Islamic knowledge systems and biomedical knowledge systems imported into the United Arab Emirates. It presents vignettes collected from interviews and participant observation in Dubai as part of nearly four years of ethnographic research, completed as part of the author's doctoral work on 'Anxiety and Identity in Southeast Arabia'. Rather than radically informing health seeking behaviours among many UAE citizens, the emphasis on the 'Arab Genome' has instead reconfirmed the authority of Bedouin cosmological understandings of disease, reshaping the language that people use to engage with their bodies and their health. Local cosmology remains a powerful discursive element that often operates in contention, in sometimes powerfully subtle ways, with novel health initiative regimes. For many people in the region, genomic information, as it is often discussed and propagated in the UAE, shares an intimate relationship with ideas of fate and national identity, and sometimes serves to mitigate the increasingly uncertain terms of engagement that people share between the body, their health, and rapidly changing urban landscapes.

Genomics and cure: understanding narratives of patients with Duchenne muscular dystrophy in Japan
Masae Kato

Globally, genomics research is expected to enhance the health of patients with intractable diseases such as Duchenne muscular dystrophy (DMD). But how do patients perceive medical and scientific attempts at creating drugs and finding cure, and why?

Since the 1990s, a number of clinical trials for patients of DMD have been organized. Among them are a gene therapy and exon skipping, and they indicate the possibility of finding therapies for DMD patients. Since 2011, Japanese medical institutions have been participating in Global Clinical Trials so that Japanese DMD patients can have access to them once developed.

Despite ongoing global clinical trials, however, field research shows that the DMD patients the author encountered were neither enthusiastic nor well informed about gene therapies developed in Japan or elsewhere. Why not? The author observed that the desire for a cure among DMD patients is not self-evident, but is framed by sociocultural conditions surrounding the patients, the local history of discrimination against genetic disorders, and the way care is organised. These factors further interplay with physical and mental conditions particular to DMD, affecting patients' desire for a cure. This paper discusses the perception of genomics research and the possibility of a cure of DMD patients the author encountered in Japan, indicating that such perceptions are a result of the deeply-related interactions of the conditions in which patients live. Finally, the author suggests how commonly held views of patients and patients' desire for cure need to be nuanced in genomic medicine.

Data in this paper were collected between April and July 2014, and between 1996 and 2005 in Japan.

The justification of studies in genetic epidemiology – political scaling in China Medical City (*open access*)
Margaret Sleeboom-Faulkner

Genetic epidemiology examines the role of genetic factors in determining health and disease in families and in populations to help addressing health problems in a responsible manner. This paper uses a case study of genetic epidemiology in Taizhou, China, to explore ways in which anthropology can contribute to the validation of studies in genetic epidemiology. It does so, first, by identifying potential overgeneralizations of data, often due to mismatching scale and, second, by examining it's embedding in political, historical and local contexts. The example of the longitudinal cohort study in Taizhou illustrates dimensions of such 'political scaling'.

Political scaling is a notion used here to refer to the effects of scaling

biases in relation to the justification of research in terms of relevance, reach and research ethics. The justification of a project on genetic epidemiology involves presenting a maximum of benefits and a minimum of burden for the population. To facilitate the delineation of political scaling, an analytical distinction between donating and benefiting communities was made using the notions of ‘scaling of relevance’, ‘scaling of reach’ and ‘scaling of ethics’. Political scaling results at least partly from factors external to research. By situating political scaling in the context of historical, political and local discourses, anthropologists can play a complementary role in genetic epidemiology.