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## Introduction

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Predictive genetic testing technology is still very much in its infancy in Western healthcare systems. However, as geneticists continue to establish links between the location of genes and particular disease aetiology, so further scientific knowledge may occasion more encompassing social definitions of who legitimately can be classified as ‘pre-ill’ or ‘pre-symptomatic’. Potentially all of us may be transformed into ‘genetic citizens’ with one kind or another of genetic ‘profile’, either before birth or sometime during our life course. But what exactly does it mean to be classified as a person with a predisposition to illness and how are the life sciences and technologies creating pre-symptomatic persons as new forms of social value?

This book is a critical exploration of the emerging pre-emptive cultures that shape the new predictive genetics. Based on original materials from fieldwork in contemporary Britain, it argues there is a pressing need for the social sciences to analyse conceptually, empirically and pragmatically how we think through the links that bind together the ideals of prophecy and health in such predictive contexts.

The ethical controversies surrounding genetic testing have largely emerged since the development of tests based on the direct analysis of a person’s DNA. This has only been possible since the identification of bio-molecular markers enabled geneticists to begin the work of tracing correlations between particular disease-causing agents and specific genes. Though successful linkage applies still mainly to the ‘single-gene’ disorders whose genetic mutations are considerably simpler to study than the more common polygenic conditions, scientific understanding of the nature of multiple interactions between different sets of genes in disease formation is commonly heralded as the next genetics ‘revolution’. The possibility for genetic diagnosis itself, though, is not entirely new. Antenatal testing for chromosomal abnormalities such as Down’s

syndrome has been offered routinely to older pregnant women and genetic screening already has some routine applications. In the UK, all newborn babies are screened for phenylketonuria, a genetic condition that can lead to serious learning difficulties unless counteracted by a special diet. For babies that test positively, the adverse effects can be pre-empted by early treatment.

It is this kind of example about the merits of early illness prevention and treatment that underpins rationalisations for the promise of a ‘golden age’ of new predictive healthcare. These rationalisations are underwritten by many of the late-capitalist economies of the West that aim to link advances in future health provision with supremacist ideas of cultural progress and power. The biosciences and life technologies are endowed in many of these visions with an implicit civilising mission. Britain, for example, aspires ‘to lead the world in the discovery and realisation of the maximum benefits of genetics in healthcare’, with the British government pledged to invest £50 million in genetic research, genetics-based health services and professional training between 2003–6, with further funding to follow (Department of Health 2003:8). Elevating in this way the gene to the newly enhanced status of visible cultural icon, it is only by appreciating the wider social implications of the predictive testing era that the claims of the original guiding promise will be open to critical scrutiny and ongoing evaluation (see figure page 3).

Taking a strong integrative approach that draws out some of the possibilities for a productive synthesis between social anthropology, cultural analysis and a critical bioethics, *Narrating the New Predictive Genetics* introduces a number of important empirical findings that extend the parameters of existing critiques of ‘geneticisation’ in significant new directions. The aim here is to contribute to a growing social science scholarship on the anthropology, sociology and psychology of the new genetics (e.g., Rabinow 1999; Conrad and Gabe 1999; Marteau and Richards 1996) by paying attention to how we formulate questions about the meaning of predictive genetic knowledge for definitions of society. Both the anthropology of biomedicine and the cultural analysis of new genetic technologies are relatively recent topics within the social sciences. A few anthropologists, for instance, have turned their attention to women’s experiences of prenatal screening techniques as well as interpretations of risk amongst those undertaking predispositional screening for breast cancer (Rapp 1999; Finkler 2000; Lock 1998; for other monographs addressing the new genetics see Rabinow 1996, 1999; Fujimura 1996; see also Franklin and Lock 2003). However there has been no critical study devoted to the shift from treatment to prevention-based medicines, and in particular no anthropological study exploring how the making of the ‘pre-symptomatic person’ reconfigures current definitions of sociality and social identity in complex, technologically



Gee! Gnomes!

"And **thus** we can see how you were **born**, how you **live** and how you'll **die**!"

12 February 2001

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*This political cartoon uses the 'breakthrough' of the first so-called 'rough draft' of the sequenced human genome to illustrate the potentially determinist reasoning behind predictive claims to genetic supremacy. In this case, a satirical play on the nature of political power depicts the internal rivalry between two government figures from the current Labour Party in Britain: the Chancellor of the Exchequer, Mr Gordon Brown, is shown finally to supersede the Prime Minister, Mr Tony Blair whose genome comically reveals next to no genes.*

advanced societies. This is a somewhat strange omission, for ever since E. E. Evans-Pritchard's (1937) seminal *Witchcraft, Oracles, and Magic among the Azande*, social anthropologists have attended to the many ways that divinatory knowledge across non-Western cultures is believed to have transformative effect through the medium of manipulated human bodies and other ritualised objects. Revisiting then something of an 'old' anthropological interest, this book offers a critical commentary on the new oracular predispositional 'truths' of twenty-first century prophetic biology and the relation of these truths to changing popular conceptions of persons, bodies and notions of genetic inheritance in biomedical Britain today.

By way of detailed case studies of families affected by Huntington's Disease (HD) – a monogenic (single-gene) inherited and late-onset condition for which there is presently no known cure – we will examine how the exchange of genetic information between kin entails unresolved processes of moral decision-making within and across the generations. Understanding, however, why such local moralities of information disclosure generate dilemmas over what knowledge is 'good' to know and what knowledge is 'bad' to tell and share with others, raises questions of wide relevance beyond the specifics of HD cases and subjective illness experiences. To date, anthropologically informed commentaries of the new medical technologies have largely neglected the conceptual question of how, and to what extent, the choices informing people's reproductive and genetic decision-making comprise so-called 'ethnographies of morality' (Howell 1997). As a consequence, anthropologists interested in this area have tended to avoid asking how, and indeed how adequately, their conceptual apparatus can address the working premises of mainstream Western bioethics. In the context of predictive genetic testing technology where consanguineal ('blood') kin who have chosen not to get tested may find another's test result implicates their own health status, such issues become especially germane. In the light of these difficult disclosure dilemmas, this study reconsiders the conceptual premises of individual autonomy informing the 'right to know' debates of contemporary Western bioethics. It finds the interrelatedness of interests informing local practices particularly suggestive for the conceptualisation of a 'genealogical ethics', which in turn may be seen as part of a wider relational amalgam (a 'relational ethics'). Additionally, I have wanted to introduce certain cross-cultural data from the existing medical anthropological record to show why such materials are salient to the wider discussion of human embodiment and identity in the genome era. Since the inclusion of comparative data has been noticeably absent from previous ethnographies of the new medical technologies, such comparisons hopefully yield additional interest and broaden the terms of debate, for anthropologists and non-anthropologists alike.

In the course of researching this book, I have lost track of the number of times people have asked questions about my intellectual allegiances. For whom does one write? To whom is one talking? For all authors, these are important, inescapable questions. As indicated, the following pages are attuned to particular anthropological sensibilities, however I want to stress that the book is written at the same time with a broader non-anthropological readership in mind. Indeed one main aim is to bring together the usually disparate domains of ethics, ethnography and science as the beginning of a critical exploration in interdisciplinary dialogue between medical and non-medical practitioners. During their daily rounds, clinical geneticists, genetic counsellors and academic

bioethicists usually do not ‘talk with’ social scientists. Just the same might be said of the latter: social scientists rarely find themselves positioned as well-integrated or long-term fixtures within mainstream scientific or medical research communities. Issues of access to clinical settings by non-clinicians are often the first impediment to such cross-dialogue. But these disciplinary and inter-institutional ‘gaps’ between the different practices and scholarly communities seem, if anything, more essential to address now as shifts towards so-called ‘Mode-2’ distributed knowledge production demand new transparency and participatory structures.

When an ethnographer chooses to work with and through certain publics – when he or she purposely mediates the creative space of the ‘agora’ (Nowotny *et al.* 2002) – then engaging the professional interest of scientists, clinical geneticists and other health professionals seriously matters. Let me be specific. Since the rate of uptake of predictive genetic testing has been far lower amongst the HD community than was originally expected by clinicians, there is a dearth of social knowledge relating to the real life experiences of genetically predisposed (i.e., ‘at risk’) but untested individuals. There is also very little public awareness of what it means for affected families and individuals to live with a ‘pre-symptomatic’ diagnosis. Living life pre-symptomatically is a skill few of us might have heard about at the present time. Indeed, clinicians themselves have cited evidence suggesting that those who experience the greatest difficulty in coping with an adverse test result are also the likeliest client group to drop out of clinical follow-up studies. Similarly, although policy specialists often pay lip service to the ‘ethical dilemmas’ of predictive genetic testing technology, the normative formulation of bioethical statements on predictive testing by various expert committees has been delimited extremely narrowly. Across Euro-America, relevant ethical bodies have not to date focused on broad inclusive questions such as how revised diagnostic tools in clinical genetics are creating ‘pre-symptomatic’ persons as new social identities. In the media too, there has been next to no debate addressing how the effects of these genetic testing technologies are creating new prognostic moralities of ‘foreknowledge’ at the level of ordinary lived experience. Based on the ‘expert’ accounts of those who have tested positively as well as those receiving good news, this book by contrast reorients the focus through illustrative examples and stories from specific contexts. With its close attention to narrativisation and issues of temporality it hopes to supplement the quantitative research which clinicians routinely consult and analyse.

## PART I

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### Ethnography as linkage map

## 1

## Thinking futures

## Lives to come

In 1994 the late French novelist and literary translator Elizabeth Gille published a remarkable autobiography about temporal dating and anticipated death. Realising her diagnosis of terminal cancer leaves limited creative writing time, Gille pens *Le Crabe sur la Banquette Arrière*, the story of a counterpart heroine who tries to put her remaining days of declining health to the back of her mind. Friends, colleagues, family and even strangers all have other plans, however. Meaning well, they rally round this ‘sick’ relation offering clippings from popular magazines on the latest ‘miracle remedies’; or they collect groceries suggesting she eat a ‘healthy’ fish diet, cook these recipes, do those exercises and so on. To her frustration Gille’s heroine is reminded continuously by others how her designated sickness role, as enforced regimen of care, predates her impending death. As the author herself remarked in the advanced stages of her illness, these kinship relations are however misplaced conceptions. ‘The date of your deaths remains uncertain, but mine is already set, more or less’, she told close friends. ‘That does not prevent me from living. Or from laughing’.<sup>1</sup>

It is no accident that the recent ascendancy of new genetic testing technologies primarily in the wealthiest markets of the late industrialised world has spawned both a sceptical and optimistic literature about the ‘dream’ of the human genome and of future ‘lives to come’ (see Lewontin 2000; Kitcher 1996). Scientists, the media, industry, bio-pharmaceutical companies each have various ‘stories’ to tell and venture interests to perpetrate about the intended benefits derived from the future creation of supposedly healthier populations. In its extreme version the vision anticipates a new era of cheap rapid genetic screening with technologies such as the DNA chip and personalised sequencing. Go to your primary care practitioner and theoretically he or she will be able to predict the probability of your getting any number of known genetic

diseases, including the common multi-factorial conditions such as heart disease, cancer and diabetes. On this basis, one's doctor could hope to recommend preventive measures before certain symptoms appear. You might be advised to come for regular check-ups, modify your diet, quit smoking, take more exercise, avoid environmental toxins and so on. Alternatively, the genetic consumer might bypass altogether the medical specialist and simply go to the local pharmacy instead. Just as 'do-it-yourself' DNA testing kits are appearing already on the market today – sold 'over-the-counter', available via the Internet or through alternative practitioners (e.g. dieticians, complementary therapists) – so in the future one might purchase one's own DNA sequence directly as a disk to self-analyse at home on one's personal computer.<sup>2</sup>

But would we all live longer, healthier and happier lives as a result? For the major pharmaceutical and biotechnology companies, the question may be tangential to other prime considerations. Namely, the perceived benefits of pre-dispositional profiling turn in part on the generation of near-term revenue and the return of pharmaceutical profit for previously patented genes. The expansion of the drug market to 'pills for the healthy ill' may also precipitate onerous forms of commercial and psychological exploitation through the manipulative 'marketing of fear'; something of an antidote to the calculations of pharmacogenetics and pre-emptively tailored individual drug responses (Gilham and Rowland 2001; Moynihan *et al.* 2002; see also Davison *et al.* 1994). Such concerns tend to be countered in existing policy debate by the presumption of the active information-seeking subject and the belief that expected benefits for the populace at large turn on the individual's supposedly free choice to make responsible genetic interventions to stave off disease – this especially so against an ideological backdrop of advanced liberalism and active citizenship (for sociological critiques see Novas and Rose 2000; Koch 1999). Across these concerns one hears some research geneticists articulating the intellectual caution brake. Apparently doubtful of the predictive power of genetic medicine for the treatment of polygenic complex disorders, such developments – it is claimed – are at least some twenty to thirty years away. Of course such doubt may serve at times as another promotional strategy: the scientists' assuagement of the public's confidence. A recent refrain at academic conferences and 'science and society' events goes along the lines: 'Don't worry – things aren't running out of control – the complexity of risk quantification for common disorders is way beyond [even] us!' Meanwhile, the goal of developing a radical breakthrough (in terms of cost and throughput) in sequencing of genomic DNA has been captured in the slogan 'the thousand dollar genome' (i.e. sequencing the whole genome of an individual for about \$1k in about a day). This was first



articulated at a ‘visionary meeting’ organised by the National Human Genome Research Institute (NHGRI) and chaired by Francis Collins in 2002.<sup>3</sup> In conveying these various rationalisations to the public, certain sectors of the media tend to muddle and simplify the picture with inaccurate reportage of scientists finding genes ‘for’ certain conditions, as if genomic-based science were fundamentally a matter of straightforward causal correlation between a gene and the phenotype, to say nothing of differences in social structure, lifestyle and environment. Through these overlapping and oft contradictory claims ‘Biotechnology’ mixes benefits with harms in one seamless package such that the knowledge outcomes of the Human Genome Project often collide in a supercharged vacuum of gung-ho determinist triumphacy. A collision that anthropologist Paul Rabinow (1999:23) derides as the ‘hyperbolic discursive tidal wave of hope, fear and metadiscourse’, and one that some clinically trained practitioners condemn with equal opprobrium as the dangers of a new age of medicalisation and rhetorical hype (Holtzman and Marteau 2000; see also Melzer and Zimmern 2002).

It is precisely such sensationalism strategies that I want to move away from so as to reorient debate through a different analytic trajectory. By traversing the conventionally discrete domains of ethics and science in post-Enlightenment European philosophy, this book unfolds as a cultural exploration of the way ethnographic analysis can be deployed as a critical tool to mediate the worlds of objective scientific ‘fact’ and subjective ethical ‘value’. Part 1, ‘Ethnography as linkage map’, outlines some key themes and locates the nature of the ethnographic problem in terms of a culturally resonant ‘linkage map’. Before I start to sketch in these points of linkage, let me account a little more explicitly for some of the ethnographer’s own concerns. Social scientists may be trained to deride hype, but such critical detachment does not abstain me from participatory engagement, albeit more subtle and reflexive forms of involvement. Nobody after all can write as though they were *tabula rasa*.

I want to present three caveats along the way. The first is nothing more than an acknowledgement. It is to make the rather simple but critical point that a wide range of genetic tests with different degrees of predictability is currently under development. It is then seriously misleading to talk about predictive medicine as though it were a monolithic enterprise, since in so doing we underplay the significant difference between those high-risk families with a known hereditary illness (single-gene inherited diseases) and common complex diseases in the wider population (Mathew 2001). For the latter, the presence of gene variations or ‘polymorphisms’ may mean that genes represent fairly poor predictors of

disease. Take the example of workplace hazards and the case of an employer wishing to test job applicants with a predictive susceptibility test prior to the offer of an employment contract. Now a person's potential susceptibility to a chemical could be affected by hundreds of different genes that encode enzymes and molecules involved in many different metabolic pathways. Rather than any single genetic difference it may be the overall pattern of gene variation that will influence the possible onset of a health problem. Or genetic differences may be attributable to different metabolic transfer rates whose effects cannot be easily predicted. You may be able to break down toxic chemicals efficiently that prevent the development of a predispositional risk factor, whereas my body might not produce the right level of enzymes in the right amount, even though I feel and appear quite healthy. If we both keep our distance from the group of chemicals known as arylamines (associated with dyes, textiles and rubber manufacture), then theoretically our different metabolic rates as fast and slow 'acetylators' will be of negligible predictive value for the NAT2 gene variation linked with the increased susceptibility risk to bladder cancer. But the added caveat reveals the complex subtleties at work. The genetic variation in NAT2 that is thought to increase the risk of bladder cancer is also thought to reduce the risk of developing colon cancer. All in all, I may be more protected from colon disease than you! Any predictive genetic test result could therefore involve the misinterpretation of an individual's actual risk, thereby leading to social inequality through practices of genetic discrimination – my not getting the job appointment, for example.<sup>4</sup>

Second, as more tests for multi-factorial genetic disorders become available in the coming years in the form of so-called 'pre-dispositional' diagnostics, we need to think much more carefully about what is meant by the umbrella term 'preventive health'. This is especially so since preventive genetic medicine is couched so often in terms of helpful treatments and effective care, omitting to say that health prevention as a practice and ideology is also tied up closely with the political economy of health systems. If health policy administrators keep an interested eye on developments in the new life technologies, this is partly because it will be more cost effective to 'screen out' persons preconceptively or to treat certain conditions prophylactically, than it will be to subsidise the cost of long-term care for those with chronic symptoms. The National Institute for Clinical Excellence (NICE) produces for the National Health Service in England and Wales authoritative guidance on the clinical and cost effectiveness of healthcare interventions and on the treatment of clinical conditions. NICE has already produced clinical guidelines in familial breast cancer and undertaken appraisals for two medicinal products, trastuzumab (Herceptin) and imatinib (Gleevec) that require the prior genetic analysis of tumour cells