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Book Review

Frameworks of Choice: Predictive and genetic testing in Asia
Margaret Sleeboom-Faulkner (ed.)
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This edited volume offers a rich collection of case studies on predictive and genetic testing in Asia. Referring to the growing body of bio-ethical scholarship in Asian countries, editor Margaret Sleeboom-Faulkner argues that this book makes a case for a social science approach. This, she suggests, would allow for a better understanding of the context and implications of new genetic technologies and would put bioethical reasoning into perspective. For the theoretical analysis of the findings some key concepts are introduced, such as *frameworks of choice,* which draws attention to the circumstances and socio-economic background delimiting and conditioning the choices of individuals, families and communities. How people deal with their choices in the light of their experiences and their ways of coping with disease is referred to as *ramifications of choice*.

The chosen format will appeal to a wide audience because each chapter addresses different aspects and examples of genetic testing and screening in countries as diverse as Sri Lanka, India, Japan and China. The accounts, which are based on original field work or interviews, draw upon a range of cultural, economic, political and historical factors relevant for genetic testing. In the concluding chapter the editor summarises overarching themes and discusses general trends, such as the rise of commercial services and direct-to-consumer genetic testing. In the aforementioned countries, with the exception of Japan, genetic services are not widely available. In larger cities, hospitals and sometimes commercial enterprises offer genetic services and counselling that may not be available for the majority of the urban and rural population because they are too expensive or too far away. If genetic services are available, often infrastructure or funding for follow-up and treatment or interventions are lacking.

The 'therapeutic gap' is, for instance, discussed by Simpson in a chapter on screening for Down syndrome in Sri Lanka. In this context this term refers to the lack of intervention available after testing, because abortion is officially forbidden in Sri Lanka. In some cases women may obtain illegal abortions after an unfavourable test result. In Kumar Patra and Sleeboom-Faulkner's account of genetic screening for sickle cell disease in rural and tribal communities in India, the therapeutic gap becomes painfully clear. Sickle cell disease is a haematological disorder that does not raise problems in carriers (heterozygotes), but children of two carriers have a one in four chance of developing the disorder. Kumar Patra and Sleeboom-Faulkner discuss several screening programmes that are offered by state or private initiative, including charities. People that come to the hospital or screening centre are tested and periodic screening camps can be organised in a region, sometimes in combination with health check- ups. In some cases whole villages are tested after agreement with the village community leaders. But in cases where test results show that a person has the disorder, treatment is not always available. Test results may be communicated in ways that allow other villagers to learn each other's carrier status. A host of ethical issues arise: privacy and informed consent are not guaranteed; the test result may be misunderstood by confusing

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carrier status with having the disorder; and carriers may be stigmatised, making it difficult for them to find a marriage partner.

Discrimination and stigma are a relevant theme in many accounts. Having a disabled child or family member may have serious consequences for other family members. In a chapter on testing for Duchenne muscular dystrophy, an untreatable childhood disorder, Sui and Sleeboom-Faulkner discuss how this affects genetic counselling in China. Genetic counsellors initially avoid being too explicit in explaining the hereditary character of the condition. Although the mutation can occur spontaneously, in two-thirds of cases it is passed on via the mother. Counsellors fear that the family of the mother may be held 'responsible' for passing on the disorder. If mothers were to be abandoned after a positive test result, their prospects would be grim.

Several authors discuss how religious beliefs may 'explain' a genetic condition, for instance the belief that a person or a family must have done something wrong in a previous life. Though this may further stigmatise those with the condition, they sometimes seem to express acceptance of it as being something they 'need' to experience. It would be interesting to compare these concepts with Western accounts of having a disabled child or family member. Although religious frameworks may differ, concepts such as destiny, fate and the notion of being 'tested' may show convergences in relation to coping strategies.

Kato discusses the complex context of prenatal testing in Japan. On the one hand, discrimination and stigmatisation occur when people have a handicapped child, which may lead to a greater uptake in prenatal testing. On the other hand, there is a strong coalition of feminists and the disabled rights movement opposing prenatal screening and testing as a form of eugenics. As Tsuge discusses in a chapter on experiences of prenatal testing in Japan, this opposition is grounded in experiences under the Eugenic Protection Law in force between 1948 and 1996, which limited the rights of disabled people to have children and allowed abortion of affected foetuses. Given the sensitivities in Japanese society regarding these recent eugenic policies, doctors are sometimes reluctant to inform pregnant women or prospective parents about prenatal testing nor, as far as maternal serum screening is concerned, are doctors required to inform them, thereby in fact lowering public knowledge and the uptake of prenatal tests. Kato suggests that it may also be the case that the whole idea of testing is more or less ignored by prospective parents as a strategy expressing faith in the strength and health of the family (including forefathers) and avoiding being associated with having bad genes.

The issue of free or autonomous choice, the cornerstone of genetic services in the West, surfaces in many chapters of this book. Raising a handicapped child is costly because of expenses for treatment and because it reduces the amount of time parents, particularly mothers, may spend earning a living. Families in Asia are often closely knit and for financial reasons as well as to avoid stigmatisation parents, parents-in-law or other family members may exert pressure to opt for predictive genetic testing for a late-onset disorder, or prenatal testing and abortion in case of an affected foetus. This is evident in the chapter by Gupta on genetic testing and screening in India and in the chapter by Saxena et al on genetic services in the same country. In the latter account it becomes clear that standards for quality assurance, such as laboratory standards, as well as guidelines for predictive testing do not always function. For instance, when a couple travels a long distance to the clinic, there simply is not

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much time for reflection between an initial counselling session and the performance of a test, as is recommended and standard procedure in many countries.

The issue of free choice is also prominent in Döring's chapter on prenatal sex selection in China and the abortion of female foetuses. The Chinese government strictly prohibits prenatal sex selection, in this case limiting free choice from the best of intentions. The alleged free choice considering this and related practices such as female infanticide and neglect of girls itself should be considered in a historical and social context, as Döring shows. The preference for males, especially in rural areas, is attributed to their qualification to continue the family lineage and their labour power. This is significant because in the 1980s the system of rural cooperatives and healthcare infrastructure at the village level collapsed. Though the consequences of these developments are not explained in detail, this probably made living conditions in rural areas more difficult. At the same time technological innovations such as ultrasound became available. The one-child policy had a further impact. In rural areas couples eventually were allowed to have a second child in case the first one was a daughter, but then a second daughter would be regarded as less desirable. It would have been interesting to learn more about the micro level of Chinese couples and health care workers performing prenatal screening, especially since Döring suggests there are signs that a family of four consisting of parents, son and daughter is becoming a preferred model. In some other chapters interviews work well in illustrating the arguments people use to justify their choices in a particular situation.

The role of the state, for instance in establishing regulatory frameworks, is explored in Porter's chapter discussing the lack of regulation for insurance companies in Japan on how to deal with genetic test results. Clients of medical insurance companies may be held responsible if they fall ill with or are diagnosed with a genetic disorder after the start of their contract. One of the drawbacks, which has also been observed in Western countries, is that people may not undergo testing out of fear of losing their insurance. Porter argues for a fair balancing of the interest of insurance companies to apply risk selection with the interest of the public in obtaining life and health insurance.

Market forces also feature in a chapter by Wallace on direct-to-consumer nutrigenomic testing. The idea is to test a person's genes in order to give tailored dietary advice or prescribe dietary supplements. However the validity of this advice is questionable, and may needlessly alarm people that they need certain food supplements to prevent illness. The relation between genetic variants and common disorders is complex. Genetic and environmental factors interact, and food metabolism only adds to this complexity. Wallace notes that the marketing of nutrigenenomic tests is seen by researchers in the field as premature at best. Another prominent concern raised in this chapter is that the research focus on nutrigenomics and the development of perhaps useless tests is not contributing in any way to the real problems of malnutrition and lack of food across large parts of Asia, and has a potential to undermine healthy eating messages.

A similar perspective examining globalising political and commercial forces is present in a chapter by Lee on pharmacogenomic research in different populations. There is at least some scientific evidence that genetic variation may be associated with response to certain drugs. Much research has focused on so-called slow versus rapid metabolisers, implying that the dosage of certain drugs may be adjusted to a person's individual genetic make-up. In some

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cases an alternative drug might be prescribed in the presence or absence of a particular genetic variant. Although the aim is personalised medicine, an important research strategy is to detect genetic variation in populations. As Lee argues, the International HapMap Project included individuals of Chinese, Japanese and north European descent, and Yoruba from Africa. Genetically speaking the Chinese and Japanese would not differ that much. Though for scientific reasons a greater diversity might have been better, both China and Japan could contribute to the investment and infrastructure necessary for the project. In this way a model of the 'Asian' genome is produced while many other specific populations in that part of the world are excluded from the research. This also holds for subsequent efforts to find specific 'national' genomes in the most prosperous countries, such as in China, Japan and Korea, linking national identity, race and biological specificity. Lee points out problematic social ramifications, such as defining who is, for instance, genuinely Korean. But a major concern is that populations would be unable to reap the pharmacogenomic benefits from a skewed 'Asian' genome. Lee therefore suggests focusing on individual rather than population genetic variation. One could argue, however, that with the advent of increasingly cheaper next generation sequencing, by comparing individual genomes from a variety of populations, we will increasingly be able to detect relevant genetic variation that may subsequently be tested at the individual level.

This volume provides the reader with valuable, detailed and localised information on the ramifications of genetic and predictive testing in a variety of settings. At the same time it bears testimony to the fact that in many cases we are not simply dealing with dissemination of a 'Western' technology in another part of the world. On the contrary, various countries and regions in Asia have become part of the global commercial and scientific mainstream. It is telling that in March 2011 newspaper headlines reported that Chinese scientists had obtained second place in the ranking of the number of articles published in scientific journals, authoring about 10 per cent of all scientific articles. Japan holds the fourth position and India the 10th. Learning more about the everyday practice of using genetic technologies in this part of the world is essential for everyone interested in the dynamic relationship between genetics and society.

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