Chapter 2
Genomics and Population Health:
A Social Epidemiology Perspective

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ABSTRACT

Imagine being able to find out how a drug will affect you before you take it... receiving a medication that is specifically tailored to treat your disease, while minimizing your risk of developing adverse effects. Although a person's environment, diet, and general state of health can all influence how he or she responds to medicines, another important factor is genes. Pharmacogenetics is the study of how your genes affect the way your body responds to a medicine. Pharmacogenetics helps to determine what the right medicine is for you, based on your own genes.[1] The Pharmacogenetics and Pharmacogenomics Knowledge Base http://www.pharmgkb.org/resources/education/phar-genetics.jsp.

SARS (Severe Acute Respiratory Syndrome) can be contained despite the absence of robust diagnostic tests, a vaccine, or any specific treatment. When awareness, commitment, and determination are high, even such traditional control tools as isolation, contact tracing, and quarantine can be sufficiently powerful to break the chain of transmission. - Gro Harlem Brundtland Director-General, World Health Organisation. (WHO website, accessed on July 5, 2003)

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SOME PERTINENT QUESTIONS FROM THE SARS EPIDEMIC

In the SARS epidemic of 2002-2003, the microbial agent involved (SARS coronavirus) was swiftly identified and sequenced in a remarkable collaboration between otherwise highly competitive laboratories in Asia, Europe, and North America (World Health Organization, 2003).

Notwithstanding the rapid success in isolating (Peiris, Lai, and Poon, 2003) and sequencing (Ruan, et al., 2003) the SARS coronavirus, the epidemic quickly subsided in the absence of reliable diagnostics, vaccines, or efficacious therapies.

WHO gave much credit to institutional responses such as isolation, contact tracing, ring fencing, and quarantines (i.e. centuries-old techniques), with lesser mention of personal risk avoidance and the possible contributions of seasonality effects or cross-reacting immunity from related endemic micro-organisms (Ng, Turinici, and A Danchin, 2003).

Most importantly, the economic and financial stakes involved (Chan, 2003) ensured that SARS would not be a "neglected disease" [2].

A number of pertinent questions arise from these observations, which could be asked more generally of emerging biomedical technologies:

- How important are biomedical advances (incl. genomics) to population health and to patient care? What is the relative significance of genetics in the etiology (and social ecology) of health and disease?
- What are realistic expectations of the advances that genomics can contribute to disease control, diagnostic aids, and treatment? In what ways can pathogen genomics be most useful in epidemic control strategies?
- What are the likely trajectories of genomics R&D in the foreseeable future, given the current modalities for funding of biomedical research, the associated regimes of patents, intellectual property rights, and market-driven product development, and the chronically unresolved problems of neglected diseases of the poor?
- What would be an enabling environment for the realization of the useful potential of genomics, for an equitable harvest of benefits and a humane deployment of genomic technologies that can avoid the emergence of a marginalized genetic underclass and the imposition of arbitrary, constructed norms?

A partial response to the above questions was offered by Holtzman and Marteau (2000) who argued from a clinical perspective that *"the new genetics will not revolutionize the way in which common diseases are identified or prevented… only a small proportion of the population has Mendelian disorders, and this will limit the ultimate impact of the Human Genome Project. Our doubts stem from the incomplete penetrance of genotypes for common diseases, the limited ability to tailor treatment to genotypes, and the low magnitude of risks conferred by various genotypes for the population at large. Consequently, most people will have little interest in learning their genotypes."*

The completion of the sequencing of the human genome in 2000 provided the occasion for extravagant claims for genomics as an all-round panacea for the major health (and social) problems of humanity in the 21st century. Notwithstanding this *genohype*,[3] there has been limited success thus far with gene-based therapies, and few promising candidates on the horizon.[4,5,6,7] Commercial interest is thus likely to shift towards genetic testing for 'disease susceptibility' alleles in line with a 'paradigm shift' towards 'predictive medicine' (genetic profiling of individuals for assessing risk of future illnesses). This has the added attraction that mass markets are involved, since the genetic testing for 'disease susceptibility' may be applied in a routine manner as part of well-person (or well-child) care and screening. Accompanying this almost certainly will be corporate research and development (R&D) aimed at producing 'pills for the healthy ill' (the worried well)[8] to carve out sizeable new markets not just for screening tests but also for 'prophylactics' for those deemed to be 'at risk' and consequently anxious for the availability of some (commodifiable) risk reduction options. Conversely, corporate R&D will continue to ignore and bypass the "neglected diseases" of the poor, a scandalous situation which has been well documented by *Médecins Sans Frontières* (MSF).[9]