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Promising deliveries, delivering promises

The announcement of the preliminary sequencing of the human genome in 2001[1,2] stirred much hope and hype. "The book of life"[3] had been opened, and the secrets not only of pathologic processes but also of normal development were within our grasp. Certainly, it is titillating to think that common disorders like heart disease and asthma,[4] and even susceptibilities to medication-related adverse events,[5] may have genetic components that we will one day identify, and even modify.

Before the Human Genome Project, if either side held sway in the nature–nurture debate it was probably the apologists for nuture, who cultivated our understanding of the influences of socioeconomics, nutrition, education and early childhood experience. Genetic determinism — along with the eugenics movement — was swept out of fashion by faith in equal opportunity. But with the boom in molecular genetics, the "nature" argument has acquired a new lustre and sophistication: it begins to appear that we really are, with exquisite subtlety, defined by our genes. If this idea is tolerable, it is only because we are also beginning to believe that we can achieve mastery over our genetic selves with individualized diagnoses, customized drug therapies and the molecular corrections of gene therapy. This is genetic determinism with a difference.

Or is it? Have we left the spectre of eugenics behind? As many patients who donated blood for the purpose of identifying their disease's causative gene will say, they did so in the hope that results of this research would be used to help treat people like them. However, gene-based therapies usually lag significantly behind genetic diagnostics, and prenatal detection and termination of affected pregnancies are often the only medical intervention we can offer. The emergence of new genetic technologies, together with a lack of protection against genetic discrimination,[6] may only make more insidious the social and economic pressures to produce, as in the heyday of the eugenics movement, "fitter families."

DNA microarrays and other technologies[7] (see page 253) may make it possible to screen for a host of genetic deficiencies rapidly and inexpensively. One can imagine the day when individuals can send a blood sample to commercial Internet-based diagnostic services to be screened for dozens of conditions ranging from hereditary hemochromatosis to Alzheimer's disease or depression. Such tests have a great potential to make many of us (and our insurance companies, employers and governments) see ourselves not as healthy, but as not yet sick[8] (see page 275).

It is primary care physicians who, against the incursions of genetic self-awareness, will likely end up holding the fort of nurture. Their biggest challenge may be to help patients understand that genes rarely tell the whole story; for the most part, they do not sentence us to characteristics or diseases so much as predispose us to them. Until the revolution in gene-based therapeutics catches up to the diagnostic revolution, primary care physicians may have to continue doing what they have always done, which is to help patients modify those other risk factors that act in synergy with the chromosomal hand we have been dealt. — *CMAJ*

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