BOOKS & ARTS

A reality check for personalized medicine

Bringing genetic information into health care is welcome but its utility in the clinic needs to be rigorously reviewed, caution **Muin J. Khoury**, **James Evans** and **Wylie Burke**.

Personal Genomics and Personalized Medicine
by Hamid Bolouri
Imperial College Press: 2010. 280 pp. £34

Excitement about how genomics will transform health care grows every time genes for common diseases are discovered, and with each technological improvement and reduction in price. Hardly a week goes by without another distinguished scientist proclaiming that the time is ripe to reap the health benefits of the genomics revolution.

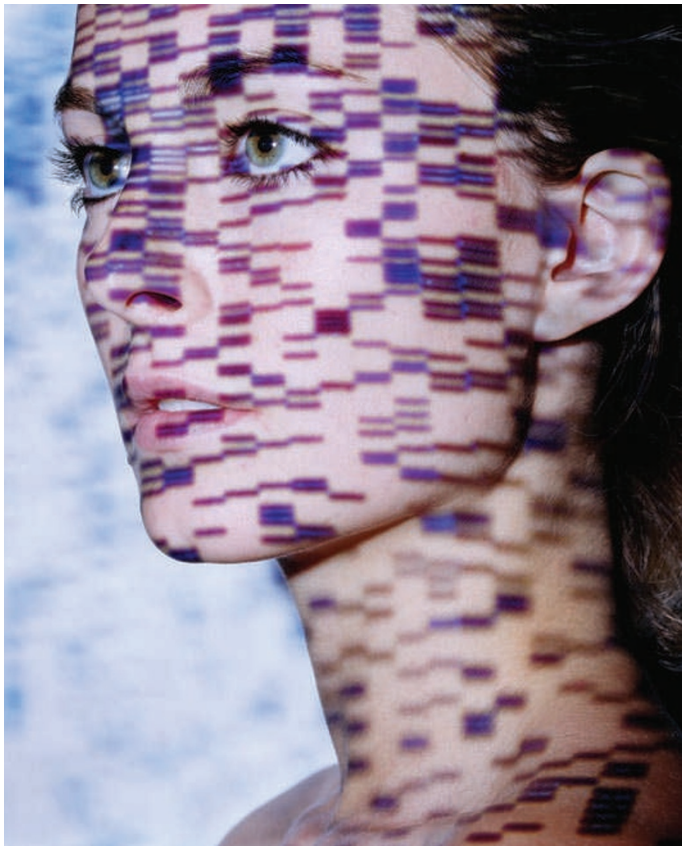
Hamid Bolouri's book *Personal Genomics and Personalized Medicine* looks at the biological, technological and methodological developments that are fuelling this growth. The author, a biologist at the California Institute of Technology in Pasadena, is a self-described "technology enthusiast" rather than a practising physician. He covers everything from DNA to physiology, including the effect of sequence variation on cellular and system biochemistry, the impact of biochemical differences on health outcomes and the intricacies of DNA sequencing and testing technologies. Importantly, he acknowledges the effects of environmental processes and gene–environment interactions on physiology and health.

Yet the unfettered enthusiasm for personal genomics and personalized medicine shown by Bolouri and authors of other recent books on the topic is not always helpful. His chapter on ethical, legal and social challenges conveys the popular notion of 'genetic exceptionalism' — the idea that genetic information should be treated more carefully than other medical information and so should be given extra legal protection. But, although it is more complicated and sensitive, genomic information is not inherently different from the results of a knee X-ray or a haemoglobin level. Only a few of the genetic tests being developed will raise exceptional privacy issues — notably tests for psychiatric or behavioural traits. There is no reason to suppose that gene variants or protein markers associated with an increased risk of heart disease, macular degeneration or a host of other common conditions will require special protection, any more than non-genetic risk factors and biochemical markers do now.

The idea that genomics information should be afforded special status has encouraged researchers to allow it to pass too easily as clinical evidence compared with other interventions. The most conspicuous example is the rapid commercialization of personal genomic tests, based on limited studies of clinical validity and none on health benefits and outcomes. Bolouri assumes that personal genomics will decrease health-care costs "as a result of better diagnostics, more effective treatments and fewer episodes of severe illness", yet out of a list of applications for personalized medicine tabulated in the book, only a few have withstood the rigour of evidence-based reviews.

Whenever new health technology is offered directly from the Internet or through clinical testing, it is tempting to think that the rules have changed. But they have not. It is important to remember that good ideas and scientific discoveries alone are insufficient to guide medical practice. This is demonstrated by recent cases of hormone-replacement therapy and prostate-cancer screening, in which the harms have often outweighed the benefits. We must hold personalized genomics to the same rigorous standards of evidence that we expect of any field.

If genomic innovations have great power to help, they also have power to harm, and patients and the public deserve a rigorous evaluation of what scientists bring to the table. It will not be easy. Genomic medicine presents profound challenges. Never before has the gap between the quantity of information and our ability to interpret it been so great. Whole-genome sequencing will produce abnormal results in all who are tested: everyone will have positive results, false positives and false negatives. Some results may prove harmful; some will be useless. Preserving the health benefits of genomics while minimizing the harms will be an important research goal. In that regard, Bolouri's exploration is a timely addition. ■



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Genomic information: should it be treated in the same way as X-ray results?

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