As a clinical geneticist, I am not too surprised that many public health practitioners appear to have ignored the well-publicized advances in genome science. Their response is understandable since Mendelian and chromosomal disorders are generally rare, while for common, complex diseases, the progressive elucidation of genetic variation has yet to impact significantly upon prevention, diagnosis or treatment. But, like Zimmern, I am concerned that the public health community needs to act now if it is to contribute to the ‘responsible and effective translation of genome-based knowledge and technologies for the benefit of population health’.

Opportunities for the judicious application of genomics are emerging across a wide range of health settings, but the potential costs of irresponsible, ineffective or inappropriate application are high. Public health will simply not be able to contribute optimally to improving health in the twenty-first century unless it factors genomic science into its practice.

There are several areas that could benefit from public health input in the short to medium term. These include the design and implementation of screening programmes that stratify the population according to genetic risk (in addition to traditional risk factors such as age); assessment of clinical utility and health economics in genetic diagnosis, including pharmacogenetics, somatic genetics (e.g. for selection of cancer therapy) and infections as well as traditional inherited disorders and the development of coherent policy and appropriate commissioning in relation to these areas. Public health practitioners could also facilitate the implementation of services that are developing or that should be developed at the interface between clinical genetics and other specialties. At present, many such developments occur ad hoc and on a limited geographical basis in response to the energy and enthusiasm of niche specialists. A notable example is familial hypercholesterolaemia, which has long been ineffectively identified and managed (at a population level) in the UK, despite the ready availability of diagnostic tests and cheap and effective treatment—a situation that should begin to change in Wales and Scotland where specific services have at last been implemented.

In order to help, public health practitioners will need to understand the power and limitations of genomic science in relation to human health. The technical and jargon-rich language that comprises the now vast biomedical literature in this area may be off-putting or even daunting to the non-geneticist. There are, however, many opportunities for learning through short courses and conferences and a number of helpful, well-researched and well-written documents relevant to public health professionals are readily available (see www.geneticseducation.nhs.uk, www.phgfoundation.org and www.sgph.org.uk). Potentially interested trainees might explore the possibility of a clinical or research placement within a clinical genetics unit while long-established practitioners and those feeling weary could perhaps be rejuvenated by a sabbatical attachment. Simply arranging a coffee with a colleague from clinical genetics could prove to be a good investment.

In his paper, Zimmern also expresses a second major concern. He feels that public health practice is stuck in an outdated and paternalistic modus operandi that fails to engage with the people it is seeking to help. He relates this divide to the growing dominance of individual autonomy in many contemporary societies (including our own). Further, he proposes that our increasing ability to describe ourselves and our susceptibility to disease in terms of genomic variation can only fuel this trend in relation to the way we use health data.
services. At present, this is probably only really true for those who may be affected by or have a family history of a genetically determined disorder. But the situation is changing fast. Plummeting costs and increasing access to comprehensive genetic sequence analysis (www.genome.gov/sequencing-costs/) will likely result in many twenty-first century citizens (and organizations) having a handle on vastly more individualized disease risk information than they do at present. Inevitably, the individuals most likely to tap into this resource will be those who have the financial and education-related resources to do so, rather than those who might have most to gain and all of us may require some protection from commercial organizations (health, food, insurance etc.) that could misuse genomics for their gain at our expense. Leadership from public health and from government will be needed.

For well over a decade, Zimmern has been challenging the clinical genetics community in the UK and more widely to recognize and embrace opportunities for working with public health. While clinical geneticists work closely with many other hospital specialities and with primary care as a consequence of clinical and laboratory referrals and the shared care of patients and families, there are few routine links with public health for many of us. Zimmern has done much to help in this regard by establishing the Society for Genomics, Policy and Public Health as a constituent group of the British Society for Human Genetics and the PHG Foundation in Cambridge as a centre of expertise in public health genetics and by catalysing international dialogue involving geneticists, public health practitioners and others. In his paper, he now challenges the public health community to reconsider the nature of its practice in the context of emergent genomic science, the changing expectations of citizens and the changing relationship between the public and commercial sectors.

References

1 Zimmern RL. Genomics and individuals in public health practice: are we luddites or can we meet the challenge? J Public Health 2011;33:477–82.