Genomics and individuals in public health practice: are we luddites or can we meet the challenge?

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Introduction

This paper is directed at my public health colleagues. It is written explicitly to challenge existing ethos and practice. It is deliberately provocative. Its message is that public health will need to amend its practice over the coming decades to take into account two important drivers to which it has so far paid little attention.

The first driver is genomic science. Knowledge derived from it has enabled us to understand with much greater precision molecular mechanisms in health and disease and has led to the development of genome-based technologies that have contributed to diagnosis, risk prediction and the management of patients. The second are the societal changes that place individual autonomy and responsibility at the heart of clinical practice, causing a retreat from professionally-led paternalism to a practice where patient autonomy lies at the heart of all health care.

My argument is that these drivers that have significantly influenced the practice of clinical medicine over the last two decades must also now lead to a change in public health practice. Some of my colleagues, particularly those in academia, are aware of and perhaps even agree with this contention. But the majority of public health professionals have shown little sign that they appreciate such trends, and the day-to-day practice of public health in the UK has not changed significantly by way of response.

Public health genomics was established in 1997 as a field of study and practice which places genome-based science at its core, and is now developing internationally into a specific and recognizable subspecialty.1 Since the meeting of experts in Bellagio in 2005,2,3 its official definition has been agreed to be:

‘the responsible and effective translation of genome-based knowledge and technologies for the benefit of population health.’

My contention is not that the existing activities of public health practice are inappropriate or unnecessary. It is that they will not be sufficient for or meet its needs in the twenty-first century, and must evolve to take into account the issues set out below.4

The impact of the genomic revolution

The completion of the Human Genome Project and the rise in post-genomic science have opened out a huge set of new possibilities for the improvement of human health. It has brought to the fore, with greater clarity than ever before, the false antithesis between social and medical models of disease; it has exposed a set of issues concerning the relationship between individuals and population and challenged the assumption of population homogeneity in epidemiological studies; and it has been in no small way responsible for the exponential growth of biological data and biomedical literature.

Social and medical models of disease

All human traits and all disease come about as a consequence of the combined effects of genetic and environmental factors. The complexity of biology is such that, even for inherited disorders such as thalassemia or cystic fibrosis, there is variation in the phenotype which in part will have environmental determinants.5 For complex disorders, such as cancer, heart disease or diabetes, the interplay between gene and environment is so interlinked that it is futile to ask whether it is nature or nurture that plays the greater or lesser part.6,7 The evidence that political, social and economic factors may determine a population’s health is indisputable.8 But, at the same time, there is now ample evidence that genetic factors play an equally important role.

No practitioner of public health should have any doubt that biological and social factors both act to determine disease risk in a population. Yet, this knowledge is hardly if
ever, reflected in day-to-day practice. The content of the recent White Paper, Healthy Lives, Healthy People, is a case in point; references to modern biology are scant, if there at all.9 Many public health professionals have a poor understanding of biological mechanisms; many appear antagonistic to the biomedical model; some seem directly opposed to it or to give the impression that social determinants of health are all that matter. The debate about inequalities makes little mention of genetic factors and appears not to recognize that the more successful we are in optimizing social and environmental causes of ill health, the greater the chance that residual inequalities will be biological in origin. Practitioners of public health must understand that biological and social models of disease are complementary rather than competing paradigms and reflect that understanding in their day-to-day practice.

Individuals and populations

The study of epidemiology has been based on an implicit assumption of population homogeneity. This assumption has served us well for over 150 years. For example, we base our interventions on our ‘understanding’ that smoking leads to lung cancer, alcohol to liver cirrhosis or overeating to obesity. But why only a relatively small proportion of smokers develop lung cancer, for example, has only recently been questioned. Our whole approach has been to apply interventions ‘to’ a population; we frame the relationship between intervention and outcome in terms of the population as a whole. In contrast, we pay scant attention to the variation in the distribution of effect size of environmental determinants among and between individuals.

The reality is that we are all different, genetically and because of our individual life experiences. The effect of interventions and the relationship between risk factors and disease will differ from one individual to another. From a theoretical and philosophical perspective, this raises questions about the nature of a population. Is it an entity in its own right or is it a set of individuals? In the early history of public health, our Victorian forefathers intervened to construct sewage systems to benefit the population as a whole. These original environmental interventions did not require any conceptualization of populations as sets of individuals. But later in time, as social models of disease and behavioural determinants of health became the focus of attention, it became clear that certain interventions could be directed only at the individuals comprising the population and not at the population itself. The concept of the population moved implicitly from that of being an entity in itself to a set of individuals.

The study of evolution also tells us that random mutations and natural selection works through individuals and not through populations. The fundamental biological unit through which interventions work must be accepted to be the individual, with the population as the derived abstraction.10

Geoffrey Rose drew attention to the need to distinguish population prevention from high-risk prevention. His approach splits the population in two, a smaller part comprising the few individuals at high risk and a much larger part the remainder.11 But the population may just as easily be segmented into several groups based on genetic or other biological risk. Different interventions, or the same intervention carried out in a different manner, may be directed at each. By segmenting populations based on genetic or biological risk, standard public health interventions can be applied differentially to each segment with more effective and efficient outcomes. Mammographic screening provides an example.12

These considerations have a bearing on the nature of epidemiological studies and their legitimate use. Classical descriptive epidemiology describes the incidence and prevalence of disease by person, time and space and is used for informing policy and the planning of health services. Analytical epidemiology elucidates the aetiological components of chronic and infectious disease, leading to better understanding of disease mechanisms and pathogenesis. However, in recent years, clinicians have used epidemiology to predict individual risk, for example, Framingham data to predict the risk of heart disease. Is such use legitimate?13,14 The calculated risk so derived is the average risk for that subpopulation. Yet, it is presented as an individual risk, and as such may only be acceptable if confidence intervals are narrow and dangerously misleading if wide.

Exponential growth in biomedical data and literature

The exponential growth in biomedical literature has been well documented. The establishment of biobanks, the advent of genome-wide association studies and the increasing use of whole-genome sequencing will lead to a plethora of data that will only be interpretable if properly analysed. Bioinformatic expertise will be essential to proper interpretation. The evidence base for the practice of modern medicine and public health will have to take account of such data. To do so will require integration with new information systems and bioinformatic expertise.15 The professional will need access to such systems as will the patient and citizen. A prime activity for public health will be to make available such information by establishing the necessary systems and using state-of-the-art technologies. To the extent that for public health, the nineteenth century was the age of the
sanitary engineer and the twentieth century of the social engineer, the twenty-first century may well see the information engineer as the key public health worker.

**Individual autonomy, clinical practice and public health**

**Autonomy and paternalism—the need for coherence**

The importance of individual autonomy and the absolute right of the patient to decide on his or her medical treatment is now regarded as an inviolable tenet of medical ethics and practice. Yet, we appear to condone a paternalistic attitude in public health practice, where not only is the provision of knowledge and advice on its own considered inadequate, but where attempts to change a citizen’s behaviour in favour of more healthy choices are thought to be acceptable and desirable. The paternalism that is found in public health practice is incompatible with existing societal trends. If we believe that citizens should be truly autonomous, the way we practice public health may need to change to empower them to embrace healthy lifestyles, rather than to coerce them to do so.

Public health has, of course, traditionally been a collective enterprise, with an emphasis on solidarity and population values. This continues to be the case for health protection activities where nationwide programmes that protect against infectious agents or environmental pollutants are both appropriate and necessary. But, in its health improvement activities, where the predominant paradigm is lifestyle change at the individual level, the contrast with individual autonomy is perhaps less justifiable, and the role of the state as paterfamilias is more questionable.

**The role of the state in changing citizen behaviour**

The Nuffield Council for Bioethics in its *Public Health: The Ethical Issues* stated unequivocally their view that

‘the central issue in public health is the extent to which it is acceptable for the state to establish policies that will influence population health.’

Activities that have been part of traditional public health practice, whether action on the external environment, for example, the provision of clean water; or the ‘indirect’ encouragement of healthy behaviours through the use of fiscal policy, advertisements and partnerships with industry are clearly legitimate activities for government action. What is more debatable is whether and to what extent the state may legitimately seek to change in a ‘direct’ way the behaviour of ‘individual’ citizens. To provide knowledge and information for citizens, to better educate them or to implement structural changes in society is one thing; to cajole or coerce them as individuals to change their behaviour is another.

It is in this context that there is now keen philosophical debate. The distinction between ‘nudging’ (increasing the chances that people act in ways that, on reflection, they would have chosen themselves) and ‘shoving’ (increasing the chances that people in ways preferred by the shower but not the shoved) becomes important and will require more detailed philosophical analysis. The Nuffield Council’s intervention ladder which moves in eight steps from doing nothing through enabling, guiding and restricting choice to its elimination makes similar points.

The argument for autonomy is one reason why we should be concerned about generalized and paternalistic health promotional interventions directed at the individual; the other is because genomic science tells us that what is right advice for one may not be so for another. The extent to which we take such variation into account needs to be balanced against the arguments for making a general case against particular environmental exposures.

**Pluralism in population health**

Public health in the nineteenth century was about the role of the state. Two sets of players predominated: physicians who practiced clinical medicine and the government which undertook public health interventions. Today, as the Acheson definition makes clear, there are many more players involved in the promotion of health and the prevention of disease; the other ‘institutions of society’ have also a significant role. Of these, we mention the media, education, law and the commercial sector. The genomic revolution has brought to the fore the importance of innovation. Industries such as biotechnology, pharmaceutical, food, telecommunications and information technology are crucial actors on the public health stage. A modern legal infrastructure that can cope with complexities related to issues such as privacy and intellectual property is part of the same equation.

The other dominant player is, of course, the citizen. The role of individuals in society can no longer be ignored. The interplay between individuals, the state, the population as a whole, industrial corporations and other institutions of society lies at the heart of how public health and population health are to be practiced in the coming decades (Fig. 1). A collectivist approach to health dictated by government may no longer be acceptable to citizen; instead, they may demand from government ways of empowering them to make their own decisions about their health.
Implications for public health practice

Introduction

The combination of issues that we set out above provides the context for thinking about why and how public health practice will need to change. The standard collectivist approach with its implied assumption that the population is a single entity on which a public health intervention is imposed will continue to be relevant for some activities, such as health protection. But genome-based science and the greater emphasis on individual autonomy must now cause us to seriously consider the need for fundamental changes to our practice, especially across the domains of health improvement and health service organization and planning. The idea that the individual should be a dominant consideration for public health may appear to some as a contradiction. But, given the rhetoric over the last decade of involving citizens and patients as partners who take personal responsibility for their own health, we have already accepted the principle that populations are, at least for some purposes, a set of individuals.

The impact of personalized medicine

These considerations provide the basis for ‘personalized medicine’, a term capable of multiple meanings, but in the context of genomics, it is the idea that medicines and health technologies can affect us in different ways and be ‘customised to each person’s specific genetic, physiological or psychological characteristics’. In some areas of practice, such as cancer medicine, the use of genomic profiling to determine the risk or prognosis of disease or the outcome of treatment is already relatively commonplace.

For public health, it is the idea that environmental factors can affect an individual’s disease risk in different ways because of genetic endowment and that disease prevention can, like treatment, also be individualized.

The Nuffield Council for Bioethics sets out three other uses of the term: the need to: (i) treat each individual as a whole person and be respectful of their particular wishes; (ii) give more responsibility for the management of health care to individuals rather than medical professionals; and (iii) provide health care as a commodity offered in response to consumer demand. The first two are probably uncontroversial. The third may appear to provide a challenge to practitioners of public health especially when considering activities such as direct to consumer testing, medical profiling and online medicine. But, notwithstanding our concern for populations and our desire for equity, reality dictates that in future years, both medicine and disease prevention will become more personalized in all four senses of the term, and consumers will take a greater role and lead significant aspects of their health-care provision.

Our role, as public health practitioners, is to embrace these changes by welcoming innovation and the personalization of health care and ensuring that they will work for the benefit of population health. The activities that will enable this are given in Table 1. In the UK, this will mean a shift of focus from working with just the NHS and public services to one that welcomes interaction between the NHS and other institutions and appreciates the synergies that can come about with an appropriate private–public mix in health service provision.

Table 1 Activities that encourage innovation and stratified health care

1. To advocate for health policies that can respond responsibly to trends in the personalization of health care
2. To establish information systems that empower consumers and citizens to take responsibility for their health, and provide to health professionals the evidence base for the efficacy of health-care interventions
3. To regulate new technologies in a manner that will both allow innovation and protect the citizen
4. To work with industry to innovate in the field of diagnostics, devices and medicines for the benefit of the population’s health

Health service capacity and organization

These issues give rise to another set of implications for public health professionals, at least in the UK, where the organization and development of health services is a key component of our work. These will impinge on the six areas given in Table 2. Each of these merits more detailed analysis. This paper cannot cover these in any detail. It can
only draw attention to the critical need to understand modern science, if only to prevent inappropriate commissioning of diagnostics and other technologies that have not been properly assessed for their clinical utility.

**Next steps**
The approach set out by the White Paper does indeed emphasize public health leadership, but by transferring health improvement functions to local government and emphasizing only partnership with social care, it will result in the exclusion of the scientific and societal trends that are the subject of this paper, and for which public health could (but does not) provide a great deal of leadership. The role of the biotechnology and pharmaceutical industries and of health-care providers and universities in the innovation agenda is as much relevant to public health as is social care. Public health genomics has realized the need to engage with the subject of this paper, and for which public health could (but does not) provide a great deal of leadership. The role of the biotechnology and pharmaceutical industries and of health-care providers and universities in the innovation agenda is as much relevant to public health as is social care.

**References**


