

## Commentary

# A new strategic phase for genomic medicine in UK health services

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### Abstract

In June 2009, the Science and Technology Committee of the UK House of Lords published a report on genomic medicine, based on expert evidence collected over an 18-month period. Crucially, the report signaled that the use of genomic medicine was at a crossroads, due to the rapid development of new technologies, and opened up opportunities across the whole of medicine and healthcare. This commentary responds to the report's call for a new health service strategy, including a new genetics White Paper from the Government, and suggests some of the important elements that need further consideration.

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### Introduction

In July 2009, the Science and Technology Committee of the House of Lords, part of the UK Parliament, published a report on genomic medicine, representing an analysis of the many scientific, clinical, commercial and regulatory issues arising from the rapid expansion in knowledge about the human genome and how it functions [1]. The report is aimed primarily at the UK government but also seeks to influence policy in the UK National Health Service (NHS), industry, the professions, research organizations and wider society. Its recommendations around translation, integration of genomics into health services and the need for continuing widespread public engagement and debate will have resonance far outside the UK. The pace of change, the report argues, requires a new strategic phase for genomics in health services. We appraise here elements of such a strategy that will be critical if the promise of improved population health is to be realized.

The inquiry took a wide view of genomic science and medicine and its report concentrated on the emerging use of genetics and genomics within medicine and how this should be strengthened. It wisely resisted the temptation to focus only on a future related to genetic susceptibility to common disease, and took a much broader scope, noting that 'it will be several years before prediction of common diseases will lead to realistic possibility of disease prevention'.

### The paradox of translational research

With respect to translation of genomic information into the clinic, the report repeats the customary, but in our view erroneous, assertion (as articulated, for example, by Cooksey [2]) that this can be achieved through translational research. Our view is that this fails to acknowledge the many steps that are involved in translation, particularly those later phases that bridge the gap between evaluation and implementation. The core themes here include: synthesis, analysis and dissemination of knowledge from a variety of relevant disciplines, including science, medicine, epidemiology, social science and the humanities (an activity that is sometimes called knowledge brokering); stakeholder dialogue and consensus building; clinical and public policy development; services review and organization; and education and training. This is a process of change management rather than research. Given the pace of technological advance, it is essential that these final phases of the translation process are explicitly recognized and adequately funded. Our own recent experience of such work in the UK in the context of inherited cardiac disease (the *Heart to Heart* report [3]) included population needs assessment, service review, formulation of service standards, service specifications and guidelines and advice for commissioners. None of this work would have attracted research funding, and yet it is absolutely necessary if innovation is to be embedded equitably into routine health services.

### Challenges for the healthcare system

Healthcare systems face massive challenges if they are to effectively respond to the scale and complexity of genomic medicine. The House of Lords report expresses concern about current inadequacies and inequities in integration of genomics into health care. Major challenges include service reconfiguration, for both clinical and laboratory services, new commissioning processes, and budget arrangements to ensure that genetic tests can be accessed from the clinical specialty where the patient is managed (such as cancer or cardiology). The report highlights that education and clinical support will be necessary to ensure appropriate

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NHS, National Health Service.

and effective use of genetics throughout mainstream medicine. Again, our experience in policy development for cardiac genetics reinforces the magnitude of such challenges. Cardiology services that properly take account of genetics are unavailable for much of the UK population. With no previous strategic planning, inherited cardiovascular disease services have developed in a piecemeal fashion promoted by enthusiastic clinicians and researchers, usually in teaching hospitals. There is limited systematic outreach of these centres of excellence to general cardiology services that, in turn, lack the knowledge to recognize and refer appropriately. Thus, major inequities arise. On a population basis, the provision and activity of inherited cardiovascular disease services varies 10- to 20-fold between populations with the more established versus the least established services [3].

### **Genetic tests and biomarkers - and how to use them**

Diagnostics, biomarkers and predictors of disease risk will be central to the future delivery of medical care. Systems will be needed to undertake their evaluation so that tests with high clinical validity and utility may be distinguished from those that serve no useful purpose. To that end, we have long advocated for the establishment of formal systems, such as those used for treatment evaluations. For example, this should include the adoption of a standardized approach to evaluation and the establishment of expert bodies with responsibility for evaluation of diagnostic tests. However, we have also noted a serious gap in availability of the necessary data and mechanisms to generate these data and place them in the public domain [4]. As with other elements of translation, there are no funding mechanisms for laboratories to generate data on clinical test performance - such as sensitivities, specificities, positive and negative predictive values - which are necessary to implement the tests within a health system. Additionally, extensive formalized support will be necessary to ensure clinicians use tests effectively, understanding their various purposes, capabilities and interpretation and communicating this for best outcomes for patients. Finally, the re-organization of pathology services along the lines advocated by Lord Carter of Coles' review [5] should be given much higher priority.

### **Priorities for the future**

Healthcare systems around the world face many challenges. Not least, in the UK, we envisage tight restraints on resources as public expenditure is reined in. There may be a temptation to think that innovation and genomics represent luxuries we can no longer afford. Deciding on priorities requires hard judgements and we suggest here some principles that might be useful.

Firstly, we should ensure that current effective innovations are translated right through to implementation on an

equitable basis for the entire population. This will require effective practical translational structures and processes as outlined above - for example, commissioners and managers of health services with sufficient understanding, expertise and time to assess and incorporate innovations into routine services.

Secondly, we must build capacity. The current lack of epidemiologists and bioinformaticians is problematic and a necessary first step would be to review current numbers in established or training posts, with a view to developing a strategy for expansion.

Thirdly, education of the health professional workforce must continue and take place at all levels. As well as the vital work in the UK of the National Genetics Education and Development Centre, which focuses on genetic skills across the health professional workforce, there is a need for organizations involved in specialist training, such as the UK Postgraduate Medical Education Board, to recognize the importance of genomics in many clinical subspecialties and to develop appropriate training programs. The Department of Health's program to develop training of healthcare scientists to interpret genetic tests in a clinical context should also be supported.

Fourthly, we should not forget the important advances that genomics could deliver in developing countries, nor should we assume that genomic medicine will be unaffordable. While developing and fine-tuning services in the comparatively well-provided UK NHS, we should also look for ways in which new technologies could provide real benefits across the globe. For example, over the next few years the PHG Foundation will be developing a 'tool-kit', through which developing countries could assess their need for genetic services in relation to birth defects and develop an outline strategy. The tool-kit will enable users to take account of epidemiological indicators of disease burden, the effectiveness and cost-effectiveness of interventions, available services, expert and lay knowledge, and ethical, social and legal implications, and will provide guidance on relevant strategic processes.

Finally, with genomics increasingly driving the vision of personalized medicine, the relationships between clinical medicine and public health, prevention and treatment, and the respective roles of the private and state sectors in the provision of both health services and research will have to be revisited. This is a report that goes to the heart of clinical service delivery, health promotion and disease prevention over the forthcoming decades, and one that government cannot afford to ignore.

### **Competing interests**

The authors declare that they have no competing interests.

### Authors' contributions

All authors were involved in discussion of the content. HB drafted the article, CW and RZ commented and all authors read and approved the final manuscript.

### Authors' information

RZ and HB are consultants in public health medicine and CW is a trained scientist with a PhD in biological chemistry. RZ is the Executive Director, HB the Programme Director and CW the Head of Science of the PHG Foundation, a multidisciplinary organization focused on achieving better health through the responsible and evidence-based application of biomedical science.

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