Will medicine be revolutionised by genomics?

R. Harris

The excellent report *Genetics and health* from the Nuffield Trust Genetics Scenario Project explores the likely effects of genetics on human health and health services, noting that 'the medicine that has been practised up to now, and the health service we have become familiar with, will undoubtedly be subject to enormous changes'.

The report's aim is to assess the impact of advances in genetics and molecular biology on the organisation, funding and provision of clinical services, on changes in clinical practice, and on the potential for disease prevention and public health action. Thus, 'The fruits of the project would be used to alert policy makers at the most senior level in the United Kingdom to what lay ahead, and to make a specific set of recommendations about what needed to be done now in order to reap the benefits of this new science for our whole population.'

The external drivers

The report first identifies external forces, called 'drivers', but which are sometimes inhibitory. 'General drivers' are difficult for policy makers to influence in any significant way but are likely, in the immediate future, to affect our lives in many ways. General drivers were identified as advances in information technology and communications, the changing role of the professional, globalisation, the impact of decisions made in Europe, environmental sustainability, human rights and demographic change. The growth of the Internet was particularly striking in the way it is used to make genome and other research findings immediately accessible, and provides patients with a mass of data, albeit of variable quality. Two 'specific drivers' were identified which determine the extent to which genetic science would be harnessed to yield benefits for human health. The public attitude to genetics, greatly influenced by media coverage, sometimes caused concern because of widespread misunderstanding, as seen in the reaction to genetically modified foods. There is also the generally poor public understanding of probabilistic data involved in interpreting risk of disease.

Ethical issues

Reference should be made to the Nuffield Council for BioEthics report dealing with genetic problems associated with the mentally ill (reviewed in JRCPI). This concluded that the same general ethical issues applied in the treatment of any patient, 'a respect for human beings and human dignity and the limitation of harm to, and suffering of all human beings'.

Dissenting views about genomic science and human health

Participants in the workshops were divided in their views on how far and how quickly genomic science would have the capacity to improve human health. The report did not consider scientific drivers in detail and avoided precise forecasts, a justified reticence as the following divergent examples demonstrate. John Bell, Nuffield Professor of Clinical Medicine, University of Oxford believes that: 'The widespread redefinition of disease through genetics will be accompanied by the use of genetics for prediction and diagnosis and to optimise treatment in most common diseases ... within the next decade [my emphasis].' In contrast, Neil A Holtzman, from Johns Hopkins Medical Institutions, believes that: 'The complexity of the genetics of common diseases casts doubt on whether accurate prediction will ever be possible.' A middle view is expressed by William Cookson, Professor of Human Genetics, Department of Clinical Medicine University of Oxford, who concluded: 'In this early stage of the search for genes underlying common disorders, it is uncertain which truly polygenic diseases will be improved in their classification by genetics. Nevertheless, diabetes and Alzheimer's disease already appear quite differently to clinicians than they did only 10 years ago ... even if most cases of disease are due to interactions between multiple polygenes of small effect, it seems inevitable that new distinct clinical entities will emerge from every disease with a substantial family component.'

In spite of this lack of consensus, one could hardly overestimate the value of an increased understanding of aetiology and pathogenesis, although improvements in treatment have lagged behind. For example, full understanding of the molecular pathology of sickle cell disease has not resulted in better management. However, the growth of classic clinical genetic services has not been prevented by poor availability of treatment for rare Mendelian disorders, and there will be increasing need for genetic interpretations for the large total number of patients with cancer, diabetes, hypertension, coronary heart disease etc. This applies especially to those with a family history, and to the minority of patients with common diseases who

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are shown to have rare Mendelian varieties. The Nuffield report concludes that 'Genetics cannot be ignored, and a modern and responsible government must prepare and plan for the challenges ahead.' A new workload will increasingly fall upon physicians as part of their routine work.

Preparing and planning for the challenges ahead

First, there is the responsibility to understand what is required of science and the trade-offs against other social values, since scientific activity, like all other social activities, takes place in a cultural, political and economic context. Second, genetics is not a monolith, but rather a sequence of overlapping areas of scientific enquiry, some of which might move forward at a different pace from others. How then might policy makers devise a policy framework that would keep pace with scientific developments, given all the uncertainties? When might we expect health services in the UK, and indeed elsewhere in the developed world, to experience the full impact of the genetics revolution? And if medical genetics empowers patients and their clinicians to predict the likely onset of common diseases, how would patients be persuaded that it was wise to adopt a preventive strategy when evidence suggests that most people choose to ignore the risks attached to certain lifestyles?

Recommendations

Physicians will be particularly interested in the report's recommendations on health and health service provision, foremost amongst which are urgent reviews of manpower and training. The needs of non-geneticist health professionals are considered a major imperative, including appropriate genetic education in the light of the new genetics as it impacts on health services. A proportion of physicians in every specialty should be trained and experienced in genetics and formally accredited as having special expertise in the subject. There was 'general agreement ... that counselling was essential for patients both before and after testing ... and that counselling had to be supportive and non-directive' but surprisingly little emphasis on ensuring the quality of genetic counselling. Already most genetic counselling is provided by non-geneticists outside genetic centres and a National Confidential Enquiry7 initiated by the Royal College of Physicians of London showed that this is often of unknown quality and unrecorded in antenatal and other hospital notes.

The importance of the development of public health genetics as a discipline and its educational implications were recognised; surely of major and growing importance when population genetic screening is possible.

The Nuffield report recommends that the NHS should develop a five-year strategy for regional genetic services. The report recognises that in the UK regional genetic services are a model for the care of patients with rare genetic disorders that is envied throughout Europe and the rest of the world. (This happy state is in considerable part the result of work initiated by the Royal College of Physicians of London14–15.) The co-ordination of clinical, laboratory and research perspectives within a single organisational structure permits a degree of coherence not often found in other specialties. The need for continuing quality assessment of the work of genetic laboratories was recognised. The strategy should ensure the validity of genetic tests before their introduction into clinical practice and take into consideration the social and ethical aspects of genetic testing, especially in relation to employment, insurance and discrimination. The Nuffield report recommends a review of the responsibilities of medical geneticists and consequent manpower requirements in the light of their increasing workload and role in education and training.

Horizon scanning

The report recommends a UK horizon scanning mechanism to monitor future developments, particularly in pharmacogenetics and in the use of genetic tests of susceptibility as a means of predicting and preventing disease. The UK should aim to establish detailed strategies on the impact of pharmacogenetics and of susceptibility testing and prevention on health services. This would include the establishment of a joint policy forum to consider and debate the impact of scientific developments in the field of genetics and their impact on medical and public health practice. The UK Government Human Genetics Commission has already established a group charged with responsibility for horizon scanning.

Perhaps it will take some time before the full importance of genomic science is realised, but according to the Nuffield Trust report, physicians will become involved on an unprecedented scale and this requires urgent reviews of manpower and training because genetics is 'the core science that underlies all modern biology and embraces the whole spectrum of health and disease'.

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