The impact of genetics on medical education and training

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This paper explores, mainly from the UK perspective, some of the issues relating to the current, and potential, impact of advances in genetics and molecular biology on the education and research training of healthcare professionals. We start by describing some of the expectations for progress in the use of genomic technologies and genetic data in healthcare delivery and the need for policy development to ensure timely translation of advances in science and technology into improved patient care. We review briefly the likely evolution of clinical genetics service provision to build the requisite scientific basis in primary care and explore how user needs could be addressed. Strategic issues for the future medical curriculum are introduced and linked with the concerns about the current status of clinical academic research. The issues for research training, career progression, nurturing of research ‘at the bedside’, definition of the research agenda and weaknesses in both academic infrastructure and support costs are reviewed in the context of the urgent imperative for medicine to harness the accelerating pace of progress in genomics.

Genomics and molecular medicine

Genomics, the study of the genetic control of body function in health and disease, has profound implications for biomedical R&D and for the future delivery of healthcare services. One expected development will be an improved understanding of the fundamental mechanisms underlying disease, with accompanying gain in the diagnosis, prevention and therapy of major diseases. The growth in genomics also promises to redefine medicine in other ways, most notably in the areas of diagnostic testing for improved disease detection, disease staging and patient stratification, plus eventual evolution of the means to identify predisposition to disease and more precise subtyping of heterogeneous diseases1.
Molecular genetics was applied initially in medicine to characterise the monogenic Mendelian disorders, but efforts have turned to the elucidation of the genetic basis of the common diseases of multigenic origin. By determining the genetic component of intractable disorders like coronary artery disease, stroke, diabetes, rheumatism and cancer, it should be possible to find out much more precisely why they are caused and the basis for the various environmental interactions which are involved. This, in turn, should revolutionise the pharmacological approach to their management and, in addition, allow public health measures to be focused more effectively on particularly susceptible subsets of the community. In addition, work on somatic cell genetics, that is genetic changes acquired during our lifetimes, promises to revolutionise the diagnosis and management of many common cancers and, may also provide invaluable information about the mechanisms of ageing. Examples of the potential for the new genetics and the underpinning role of technology development are described in detail in the Reports of the Genetics Research Advisory Group to the NHS Central Research and Development Committee and elsewhere. Genetics R&D is expected to make a major contribution in addressing many of the health targets (heart disease, stroke, cancer, mental health) identified in the recent Green Paper Our Healthier Nation.

The increased knowledge of the basic biology of disease will eventually transform medical practice. There will be a significant transition from our current reliance on the diagnosis and treatment of overt disease to a greater emphasis on the prediction and prevention of covert disease. Consideration of the role of genes in defining susceptibility to disease is, however, too narrow a perspective – and parallel progress will be needed to understand the complex interplay of gene loci that determine disease progression, disease complications and response to treatment, plus how the penetrance of risk alleles can be increased or attenuated by environmental factors.

**Emerging implications for clinical practice and training**

These advances in molecular biology will undoubtedly affect the delivery of healthcare provision, at a time when such matters are already under considerable public scrutiny. While the introduction of genetic information will have profound benefits for the health service, good clinical practice must not be allowed to suffer by adoption of an excessively mechanistic approach that ignores the clinical and social benefits of an holistic approach and provision of pastoral care. Significant changes will be necessary in the framework for healthcare
delivery, the medical curriculum and clinical research training. These elements of service provision and development are already under intense pressure. As noted by the House of Commons Science and Technology Committee Inquiry, the potential impact of R&D in genetics has been underestimated at the policy level\textsuperscript{11}. In short, the accelerating momentum of genetics in medicine will have dramatic implications for all healthcare professionals.

The impact of genetics R&D on healthcare delivery and training, and on the role of healthcare professionals in educating the public at large on the benefits of disease prediction and prevention, provides a major challenge for those charged with guiding the introduction of new and emerging medical technologies. A delicate balance must be forged between the legitimate need to control technology diffusion while research is conducted to assess clinical and cost effectiveness versus pressures for adoption created by the media, public demand, producers and the profession\textsuperscript{12}. The continuing role, and relevance, of UK national initiatives such as the Standing Group on Health Technologies, National Co-ordinating Centre for Health Technology Assessment, National Screening Committee and specific review bodies such as the Gene Therapy Advisory Committee, Advisory Committee on Genetic Testing, Human Genetics Advisory Commission need to be assessed on a continuing basis in the context of providing an integrated and coherent framework for the introduction of genetic-based products and services, and the training of healthcare professionals.

**Building clinical genetics service provision**

The organisation and manpower needs of clinical genetics services are described in the Report of the Royal College of Physicians\textsuperscript{13}. It is beyond the scope of the present paper to examine the detail of planning human resources. As the increasing application of genetics to common disorders emerges in service delivery, the role of the consultant clinical geneticist will remain important for diagnosis and counselling. However, there is an urgent imperative to ensure appropriate incorporation of conceptual and practical elements of genetics and molecular medicine in undergraduate and postgraduate education. Links with primary care\textsuperscript{3,11} and public health medicine will be fundamental in achieving full and equitable delivery of genetic services. To date, UK genetics services have been organised primarily at the local/regional level with a strong R&D interaction. However, as testing develops for the predisposition to multifactorial disorders, the volume of activity will grow such that different models of service delivery must be explored\textsuperscript{2,7}. Models that
allow both NHS and commercial development of large-scale testing with adequate quality control, legal oversight and clinical input, so as not to divorce testing from pre- and post-test counselling, need to be examined. Clarification of the primary care team approach will be of central importance in defining the demands for professional education in genetics.

The goals of medical genetics can also only be fulfilled in the context of an educated, informed public. This objective has major implications for the secondary school curriculum and for the development of ways to build public confidence in, and understanding of, the new genetics. In the absence of this informed dialogue and educational underpinning, there is likely to be both unrealistic optimism and unrealistic fear about the new technologies.

A recent comparison of the contrasting developments in clinical genetics services in the UK and US suggests that each offers special opportunities in innovation and evaluation. While a range of pilot schemes have been set up in the UK to explore how genetics services can be incorporated into mainstream clinical medicine, few have yet been fully evaluated.

**Articulating user wants**

In their analysis of the organisation of UK services, the Genetic Interest Group concluded that medical genetics was distinct from other rapidly-advancing fields in that the unit of concern was the family and not just the individual; there was a greater emphasis on preventative medicine; and outcomes are difficult to measure in traditional ways. This last feature will remain a problem – what constitutes an index of clinical effectiveness in genetics? In the absence of clear understanding of what is meant by effectiveness (and the embodiment of this understanding in education programmes), it will continue to be difficult to resolve the major problems in UK genetic services currently perceived by users:

- Access is not always consistent, or satisfactory.
- Genetic services are not always adequately linked into other medical specialities.
- There is a shortage of resource to bring new genetics-based techniques into mainstream health service provision.

Patient interest groups covering the rare genetic disorders emphasise that services should be commissioned on a regional basis, but, as genetic information becomes relevant to the common disorders, user wants will
change and the healthcare educational implications will broaden. A Citizens' Jury was recently organised to begin to define the user’s agenda of issues in genetic testing for susceptibility to common diseases. Among the training recommendations\textsuperscript{18} were:

- Planning for the future should be much higher on the agenda of policy-makers and professionals at all levels within the NHS.
- Genetics should have a more prominent part in the national curriculum and community education.
- Adequate resources should be allocated to appropriate training of all involved NHS staff.
- Basic training in genetics for NHS staff should meet nationally-agreed standards – perhaps a uniform curriculum at prequalification stage.

### Developing the medical curriculum

When considering the impact of genetics and molecular biology on the curriculum, it will be necessary to fulfil a number of challenging objectives\textsuperscript{4}: continuing education for serving professionals; the education of new professionals; information dissemination to professionals; and the R&D agenda.

In particular, in addition to considering the needs of clinical genetics as a speciality, it is also necessary to consider how the broader teaching of genetics will be organised and how it will become an integral part of all the medical specialities. We recommend the following changes:

- Inclusion of genetic principles throughout the medical school curriculum rather than treating as a separate subject in the preclinical or clinical years.
- Suitable courses to be made available as part of postgraduate training programmes for clinicians and other health workers and as a duty of all major teaching centres.
- Some component of genetics training should be included in the continuing education programmes for doctors of all specialities.

In the US, where consideration of some of these issues is more advanced, a variety of initiatives is underway. While genetics is currently taught at all US medical schools, a minimum competency is not stipulated and it is recognised that there is a serious, and growing, gap in the ability of GP family physicians to both be aware of new advances and to provide relevant consultation\textsuperscript{19}. There is a new sense of urgency to bring genetic literacy to all practising physicians (and other health professionals)\textsuperscript{20,21}. In addition to initiatives to develop the medical
school curriculum so that genetics education is fully integrated, there are many related proposals to develop Web-based resources and CD-ROM formats for continuing education programmes\textsuperscript{19,20}.

The brevity of this paper precludes a review of the other initiatives in many countries or detailed considerations of desired content, but future evolution of the medical curriculum in genetics and molecular medicine must emphasise the following:

- The importance of the new interdisciplinary framework to integrate molecular biology and clinical genetics with pathology, epidemiology and computing; the convergence of genomics and informatics in particular heralds a new era of biomedical research\textsuperscript{22}.

- The changing focus in the role of genetics not just in studying rare Mendelian diseases but encompassing the genetics of common disorders, in terms of the correlation between specific genes and disease predisposition, the definition of the molecular basis of heterogeneity in disease severity and progression, and understanding of how genetic variability can alter responsiveness to drugs and environmental factors.

- The principles and methods of genetic counselling, to address the complex range of ethical, legal and social issues posed by genetics, particularly screening of health asymptomatic individuals for predisposition to future disease\textsuperscript{23}.

- The bio-ethical dimension of genomics has evoked substantial debate\textsuperscript{15,16}. Concern about such issues as genetic discrimination, neurogenetic determinism, eugenics and germ line gene therapy and cloning have crystallised concerns within various constituencies about the pace and scale of technological change\textsuperscript{8}. Following Tomorrow's Doctors, medical ethics and law have become a core component of the medical curriculum and a recent consensus statement from the Group of Teachers of Medical Ethics and Law in UK Medical Schools\textsuperscript{24} provides a central place for the new genetics and for related issues appertaining to confidentiality, informed consent, reproduction, medical research, resource allocation, etc.

The priority, however, is not just to change the course content, albeit this is vital. What is also needed is to prepare doctors for the changing culture of the clinical transaction arising, for example, in the use of disease predisposition data by hitherto healthy individuals\textsuperscript{25}.

**Status of clinical research**

*Identifying the problems*

There is no doubt that the scale and pace of advances in genetics R&D, and the financial and organisational implications for service delivery,
will have considerable impact at all levels of medical education and research training. These increasing demands for translating advances in basic biomedical research into advances in patient care come at a time when there are substantial constraints on the resources allocated for clinical research and training. These general issues for the science-medicine interface were identified in the UK Task Force Report on Clinical Academic Careers and reviewed in the House of Lords Science and Technology Committee Inquiry. The recent increase in number of medical students proposed by the Medical Workforce Standing Advisory Committee, if funded on a marginal cost basis, will considerably increase the teaching and training burden, providing yet further disincentives to careers in clinical research.

Thus, even if the curriculum for medical education in genetics develops appropriately, will we lose the capability to train clinical researchers? These worries for genetics clinical research training are superimposed on various concerns raised by the Research Councils in their recent input to the UK Government’s Comprehensive Spending Review. For example, there is current weakness in the integration of basic medical, clinical and social sciences research. There has been a failure to develop bioinformatics and other costly advanced computational tools and infrastructure, and there is a continuing need to establish clear guidelines to support the transparent and ethical uses of genetic databases for clinical decisions.

In a recent discussion convened to address some of these issues, research and training concerns raised by the clinical academic community were reinforced by evidence from the other healthcare R&D partners. Patient interest groups, while recognising the substantial declared commitment of the NHS to clinical research, expressed dismay at an ethos of health service conformity, the apparent lack of innovation, and lack of strategy to control the variability in supply of novel research-based products and services. Medical research charities were suffering in consequence of the poor public perception of clinical research, and the pharmaceutical industry felt increasingly deterred from investing in UK clinical research. Further detail on these perspectives and on the following points can be found in the Report.

**Nurturing clinical research**

There are several issues of concern relating to sustaining a cadre of clinical researchers of international rank, how to integrate basic and clinical research, and how best to provide the training and incentives to create excellent teachers. Basic research must urgently address the needs of the post-genomic era by building interdisciplinary frameworks to link
molecular biology to physiology, pharmacology and pathology, as well as develop relevant algorithms for population genetics and the analysis of geno-phenotypic relationships in the major human diseases. But, the greatest problems in biomedical research currently are 'at the bedside'. The challenge is to create an environment to support patient-oriented research at a time of increasing workload pressures arising from service delivery, increased educational demands and resource constraints. These issues are global and analogous problems are emerging in the US with the advent of Managed Care and the accompanying financial and performance demands being placed on academic medical centres.

**Defining the research agenda**

The initial creation of the NHS R&D function was a significant and praiseworthy event. We now need to do even better and other voices should be heard in setting the agenda. New concordats might be developed to include patients, research charities and industry, and public awareness of the benefits of clinical research must be promoted.

The most effective mechanisms for organising research for those funded from the higher education budget remains an important problem. Should every medical school cover all research areas or should some specialise and others do nothing? If there is to be a coherent promotion of selectivity in research, how should the national policy be developed?

Within the overall research agenda, there are several important policy and funding issues to address as a consequence of changing styles of research. New sources of data in genetics health research are dramatically increasing in value – for example, population databases and archived issue banks. These developments raise issues of relevance to the developing curriculum on ethics and social aspects: relating to informed consent, access, confidentiality of data and intellectual property rights.

**Research infrastructure and support costs**

There are considerable strains in the UK dual funding system, as confirmed by the National Committee of Inquiry on Higher Education. Furthermore, the funding environment for bedside research is deteriorating as a consequence of the emphasis on laboratory research resulting from the last Research Assessment Exercise (RAE), and the relatively poor performance of clinical departments in the RAE. There is an urgent case to be made for increased government funding but also to
find better ways of using current resources for research and training within both the Funding Council and NHS funding streams. For example, in primary care, much of the funding supporting professional development for individuals might be better directed to practice development for primary care teams.

In an era characterised by the need for major investment to drive state-of-the-art biomedical research, it is also important to establish how best to share resources among teaching hospitals. Centralisation in multidisciplinary research centres provides effective coverage but must be progressed with care to avoid conflict with teaching priorities.

**Training medical students and research career structure**

The Task Force on Clinical Academic Careers emphasised the importance of exploring new options in research training, in particular the intercalated BSc and the combined medical PhD pathway. The decrease in funding for the intercalated BSc is worrying, although some question its value, being based, inevitably, on a didactic rather than research emphasis. The newer joint medical PhD proposal could be valuable in encouraging substantial research experience but even the well-regarded US MD-PhD programmes suffer the potential weakness of being overly laboratory-based and not inspiring the return to clinical medicine. The effective way of training most clinical researchers is to allow 3 years of adequate fellowship training following qualification and basic training; other mechanisms to provide formal postgraduate training in genomics (for example, a MSc course) for clinicians in all specialities and general practitioners should also be considered.

In addition, in spite of Department of Health efforts (for example, *A Guide to Specialist Registrar Training*), there is still uncertainty within the medical profession about the flexibility of the entry/exit points in academic career progression when embarking on professional qualifications for the royal medical colleges. This rigidity also creates problems, for example, for industry, in promoting transfer between academic and other career options. There are also problems of poor research training and career structure for academic GPs and there is a role for the other healthcare partners in primary care training.

Solutions have been proposed for some of these concerns about UK clinical research training. The creation of University Hospital NHS Trusts could provide a new vehicle with responsibility for training doctors, conducting medical research and delivering clinical care. Learning from best practice in integrating basic and clinical research across all disciplines, developing new NHS R&D strands to encompass systematised observation and appraisal, redefining units of assessment in
the RAE, ensuring greater transparency in university accounting and tracking procedures so that external funders can clearly see what their investment is buying, should all help to improve the culture of research and training in biomedicine. The recent creation of the Academy of Medical Sciences\textsuperscript{33}, a concept based in part on the model of the Institute of Medicine in the US also provides an excellent opportunity to bring together the basic and clinical biomedical sciences and the training issues. It is hoped that the initial agenda of the Academy will give priority to genetic R&D and provide impetus to addressing many of the issues for education, training and public understanding that have been raised in this article. The emergence of the new discipline of molecular medicine as part of the long-term strategy towards better health is also of great importance in the international context\textsuperscript{34}.

It has been predicted\textsuperscript{22} that the next 50 years will bring remarkable expansion in human knowledge driven in large measure by advances in genomics and informatics. Harnessing these advances to deliver better health, and ensuring that the pace of progress in biomedical research does not pose serious threat to the future competency of healthcare professionals, represent major challenges for all those concerned with education for the medical profession and the maintenance of an internationally competitive research base whose creativity can be harnessed promptly for the benefit of improved patient care.

\section*{Acknowledgement}

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\section*{References}

7 Office of Science and Technology. \textit{The Human Genome Mapping Project in the UK. Priorities and Opportunities in Genome Research}. London: HMSO, 1994
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10 Weatherall D. *The Implications of Genetic Engineering for Medical Practice.* London: Royal Society of Medicine, 1988
20 Stephenson J. As discoveries unfold, a new urgency to bring genetic literacy to physicians. *JAMA* 1997; 278: 1225–6
21 Collins FS. Preparing health professionals for the genetic revolution. *JAMA* 1997; 278: 1285–6
23 Marteau TM, Croyle RT. Psychological responses to genetic testing. *BMJ* 1998; 316: 693–6
24 Doyal L, Gillon R. Medical ethics and law as a core subject in medical education. *BMJ* 1998; 316: 1623–4
28 Beecham L. UK needs an extra 1000 doctors. *BMJ* 1997; 315: 1487