The Nuffield Trust
Genetics Scenario Project

Genetics and Health

Policy issues for genetic science and their implications for health and health services

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Foreword by John Wyn Owen CB

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Contents

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The Nuffield Trust

The Nuffield Trust for research and policy studies in health services was established by Viscount Nuffield in 1940. Today the Trust acts as an independent commentator on the UK health scene and the National Health Service. It has set out to illuminate

current issues through informed debate, meetings, and publications; and has also commissioned research and policy studies aimed at the development of policy and the improvement of health services.

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The Public Health Genetics Unit

The Public Health Genetics Unit (PHGU) was established in 1997 by the Anglia and Oxford, now the Eastern, regional office of the NHS Management Executive. It works in close collaboration with clinicians and scientists in the University of Cambridge. It is endorsed by the University's Faculty of Clinical Medicine, and is located at the Strangeways Research Laboratory as part of the University's Institute of Public Health. It is funded by the NHS through the R & D division of the regional office of the Eastern Region, and by Bedfordshire, Cambridgshire, Suffolk and Norfolk Health Authorities.

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Foreword

As the millennium approached it seemed to the Nuffield Trust that it was important to begin thinking strategically about the issues that were likely to determine the future of health in the United Kingdom and the action that would have to be taken to achieve what were decided upon as appropriate healthcare goals by the year 2015, and indeed beyond that date. These were the thoughts that led to *The Policy Future for UK Health Project* that the Trust is undertaking in collaboration with the Judge Institute of Management in Cambridge University - a project that has been led by Dr Charlotte Dargie, Nuffield Fellow in Health Policy at the Institute.

One area of enquiry that was quite deliberately excluded from that project was the remarkable developments that are taking place in the field of human genetics, and in particular their potential impact on the future provision of health services. For one thing there was already a view in some quarters that medical genetics was likely to turn many aspects of current health care upside down and that we should be anticipating nothing less than a shift in the paradigm of diagnosis and treatment that has characterised clinical practice to date to one that emphasised prediction and prevention. If one subscribed to this view of the future then the likely impact of genetics on health services was too big a subject to be accommodated easily within the Trust's *Policy Futures for UK Heath* project.

We were therefore delighted when the opportunity presented itself to collaborate with Dr Ron Zimmern and his team at the Public Health Genetics Unit in Cambridge on what soon came to be called *The Nuffield Trust Genetics Scenario Project*. No one can predict exactly what the impact of genetics on medical practice and health care is likely to be or when it will happen. Nevertheless on the available evidence its significance cannot be underestimated and we need to plan strategically for the kinds of changes that undoubtedly lie ahead. If the hoped-for destination is improved health for the whole nation then our immediate task is to discover as much about the road that leads to that destination as possible. Most importantly we must begin planning that journey as soon as possible.

This report is only a starting point. What is required next is an action plan that sets about translating our key recommendations into practice, most notably the creation of a framework in which future policy in respect of medical genetics and the provision of health services can be located. The Policy Futures exercise that the Nuffield Trust is currently conducting and which includes the Genetics Scenario Project represents an agenda for health. It promotes the idea that innovative and long-term strategic thinking is essential if we are to anticipate the impact of scientific developments on our health services and tackle the issues that will determine the future health of our population.

The Trust is grateful to Dr Ron Zimmern and his team from the PHGU for their contribution to the Genetics Scenario Project. It is greatly indebted too to the many busy and distinguished people who gave up valuable time to take part in the nine workshops. Their contribution and expert knowledge give this report its authority.

John Wyn Owen CB

London: May 2000

I

Introduction

The impact of the new genetics on existing health services in the United Kingdom has been compared to a tidal wave, a tsunami, sweeping all before it as it bursts upon the shore. A hyperbole perhaps; nevertheless 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. Even if we believe that new developments in genetic science will gradually unfold in fits and starts, and that it will be waves of greater or lesser magnitude that will ebb and flow along the beach rather than a catastrophic torrent, it is incumbent upon us to prepare for the tides that will eventually change the shape of the shoreline of the nation's health. To be forewarned is to be forearmed, in particular when people respond with a genuine understanding of what it is that lies ahead.

THE IMPACT OF GENETICS ON UK HEATH SERVICES HAS BEEN COMPARED TO A TIDAL WAVE

Some claim that that genetics will be likened to a tidal wave whose impact will change significantly health services and medical practice within the UK

WE CANNOT PREDICT THE GENETIC FUTURE BUT WE CAN PLAN FOR IT

Even if new developments in genetics unfold in fits and starts rather than as a torrent, we must prepare and plan for the future. We will otherwise be unlikely to reap the reward of better health for all that is promised by this new science.

The Public Health Genetics Unit in Cambridge owes its existence to a recognition of the changes to existing medical services and the potential for improved public health resulting from developments within the field of genetics.

Aims of the PHGU

- To keep abreast of developments in molecular and clinical genetics, and in their ethical, legal, social and public health implications
- To provide a link between academic research, clinical practice and the development of policy within the NHS for genetics and genetic services, including the implications for the funding, development, staffing, organisation and provision of those services
- To establish mechanisms for dialogue within the NHS between geneticists, physicians, public health and primary care professionals on matters related to genetics, molecular medicine and genetic services
- To provide an epidemiological and public health perspective on NHS policy development for genetic and related services, including criteria for evaluating genetic testing and genetic screening programmes

These were also changes that the Nuffield Trust had anticipated when they decided to embark upon their current project *Policy Futures for UK Health*. In the event the Trust decided that the role that medical genetics might play in that future was too great and perhaps too important a subject to be subsumed into that project. Thus the *Nuffield Trust Genetics Scenario Project* came into being as a joint project with the Public Health Genetics Unit.

The project had a single aim. It was 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.

Aim of the Nuffield Trust Genetics Scenario Project

- 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 diseaseprevention and public health action

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.

To be certain of a destination is one thing; deciding how to make the journey there quite another. It was clear from the beginning of the project that the very speed at which developments in genetics were taking place posed a challenge to any group trying to take a bearing on where to embark on that journey. It was also clear that there was a huge diversity of views about where genetic science was leading, about the pace of development and about its desirability and its impact on society.

The human genome would be finally mapped within two years or so. Individual genes were already being coaxed into revealing their particular secrets. And arguments about patenting particular discoveries and intellectual property rights were growing more heated. The international pharmaceutical industry was committed to translating the new science into medical technology, and to taking a fresh look at how diagnostics might assume an enhanced importance in the delivery of health services. National governments and trans-national bodies, the European Union and the World Health Organisation for example, had also begun to consider the ethical, legal and social implications of this remarkable advance in scientific understanding, to take note of its possible financial implications and to wonder how these developments might further increase the cost pressures that demography, public expectation and other technological advances were already placing on inadequately funded health services.

At an early stage the team that had been brought together for this joint project, now christened the *Nuffield Trust Genetics Scenario Project*, had realised that the word 'genetics' had been used in many different ways and in many contexts, and often as an umbrella to cover a number of related but nonetheless discrete areas of scientific enquiry

Some Uses of the Term Genetics

- DNA and its associated technologies
- Single gene disorders inherited in a mendelian fashion
- The genetic component of all human traits and diseases
- Familial association
- Modern biology

It has served as a synonym for biotechnology and genetic engineering, covering all aspects of DNA and associated technologies, including the production of engineered products such as human insulin and growth hormone. More specifically it has been used to refer to diseases such as cystic fibrosis or muscular dystrophy. In other instances it has been expanded to embrace the genetic component of all human disease and traits, such as height or eye colour, or as an alternative to the word "familial", to mean features that run in families. Even more generally, the word has been used as shorthand for modern biology, to encompass the understanding that all biological processes are coded for by genes. Yet scientists are critically aware that the mapping of the human genome is only the beginning, that biology involves much more than knowledge of genetic sequences and that, in what has been called the post-genomic period, biology will become more complex, the demands made upon data processing systems ever greater and more sophisticated and the path through each enquiry more crooked rather than straight.

The *Nuffield Trust Genetics Scenario Project* did not attempt to confine its discussions to any specific view of the word "genetics", and allowed as broad an approach to the term as participants wished to give it. Throughout the project it kept in mind the complexities of its usage, but permitted in equal measure specific discussion of the problems of services for single gene disorders as well as the more general issues surrounding prediction of susceptibility to common diseases.

The knowledge that single nucleotide polymorphisms (SNPs), genetic alterations in the individual bases that make up sequences of DNA, may determine susceptibility to disease has existed for many years, and has provided the scientific basis for understanding diseases such as sickle cell anaemia, which occur with increased frequency in specific population groups. However, once we possess the technology to sequence and identify them in large numbers, and to correlate them with common complex disorders, a new era in the understanding of disease susceptibility and mechanisms will unfold. We shall have a greater insight into the mechanisms of human disease processes than ever.

The course that this science may take is unpredictable. Its ability to deliver real benefits to human health is by no means certain. The reservations that surround future developments in medical genetics are many. There are optimists who maintain that medicine as it is currently practised will change completely within the next five to fifteen years, while other voices argue that very little is likely to be different from now, or that if we are anticipating revolutionary changes, the tidal wave is still a long way off below the horizon. Moreover, transforming the science of genetics into a technology that can provide useful diagnostic procedures and therapies is likely to be costly.

GENETICS MAY HELP US TOWARDS A GREATER UNDERSTANDING OF THE MECHANISMS OF HUMAN DISEASE AND A BETTER PREDICTION OF DISEASE SUSCEPTIBILITY

As our understanding of genetic and cellular mechanisms increases a new era in our ability to predict and diagnose disease will unfold, although the pace at which these risk predictions will become useful in the clinical situation is very much a matter for debate.

It is also likely that pharmaceutical interventions may be individualised to benefit only small groups of patients with particular genetic attributes. This is a very different situation from how drugs are used today where they are prescribed for whole populations in the knowledge that some individuals will suffer side effects while others will receive no kind of benefit.

Undoubtedly the ability to document the genetic structure of individuals will allow a greater understanding of disease mechanisms and a better prediction of their susceptibility to disease, but the pace at which these risk predictions will become useful in the clinical situation, or the extent to which individuals will change their behaviour or allow effective interventions to alter that risk, is very much a matter for debate.

TRANSFORMING THE SCIENCE INTO MEDICAL TECHNOLOGY WILL BE COSTLY

The new technologies associated with genetics and the increasing expectations of patients will create significant cost pressures that will have to be carefully considered in the context of an NHS with finite resources at its disposal.

Whether these developments, diagnostic or therapeutic, will be as costly to bring to the market as the products of today, or whether greater knowledge of genetic sequences will shorten development times and reduce their costs is a matter for debate. It is also uncertain if patients will necessarily demand new genetic tests and new medicines that give only marginal benefit. Our prediction is that these new technologies will give rise to significant cost pressures, not only because of past experience, but also because of pressures generated by the media and by the expectations of an ever-demanding public. The impact of these cost pressures will need to be carefully considered in the context of an NHS with finite resources at its disposal, even taking account of government's commitment to fund the NHS at new and unprecedented levels.

Many commentators have argued that in the future the organisation and delivery of health care will change radically as the current medical paradigm of diagnosis and treatment shifts to one based upon prediction and prevention. What then will be the implications of such a change for the provision of health services in terms of education and training, organisational structures, staffing, and resources? The Nuffield Trust, in collaboration with the Public Health Genetics Unit, takes the view that there has to be merit in trying to make an informed judgement on how medical genetics might develop in the future while simultaneously considering the implications of these developments for future health care provision in the United Kingdom. The challenge for the project was how to explore these developments and their implications, and then to attempt to shape them into a coherent set of recommendations for action now. The principal difficulty lay in grappling with the uncertainties that are part and parcel of contemplating the future. Put simply, it is personal opinion, best judgement and sometimes well-informed guesswork that underpin many of the predictions about the future of human genetics. There is a paucity of hard evidence from

which to proceed when planning for the future, and the evidence that can be tested is often beset with contradictions.

Moreover, political and social trends, and technological developments in other fields, will all affect, modify and determine the future impact of genetic science on health care. Globalisation and nearer home the influence of the European Union provide a backdrop against which genetics, health care, the pharmaceutical industry, intellectual property law and many other issues have to be assessed. Demographic trends too are clearly of the greatest relevance for social and health care policies, while environmental issues such as pollution, biodiversity and sustainability bear down on work in the biological sciences. Developments in information science, ranging from information technology, through knowledge management, to bio-informatics have already and will continue to influence the practice of genetic science, while telecommunications and the internet will increasingly bring about changes in the relationship between doctor and patient, and between patients and the new kinds of knowledge to which they have access.

THE GOAL OF ALL OUR EFFORTS IS THE IMPROVEMENT OF HUMAN HEALTH

We have an interest in genetic science because we believe it will enable us to improve human health by preventing disease and treating it more effectively.

We wish to state unequivocally that the goal of all our efforts is the eventual improvement of human health. If this joint project has taken a particular interest in genetic science then it is because we believe that we can benefit from a greater understanding of disease mechanisms in order to prevent disease before it has become established, or to treat it more effectively. However in focusing on genetics we do not in any way belittle the importance of environmental determinants of human health. These continue to be of inestimable importance. Genetic factors cannot be viewed in isolation from environmental ones. Each play their part in determining the health of the individual and of populations, and for populations in particular, environmental factors continue to be of greater importance and the means by which most common diseases are likely to be controlled in the foreseeable future.

ENVIRONMENTAL FACTORS CONTINUE TO BE IMPORTANT DETERMINANTS OF HEALTH AND DISEASE

Genetic cannot be viewed in isolation from environmental factors. The environment continues to be of the greatest importance, and the means by which most common diseases will be controlled in the near future

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Nor can genetic science be separated from the delivery of health services. The notion that the gene is fundamental to all cellular processes and an integral component of all human biology and pathology will ensure its importance for every aspect of health care in the years to come. A modern and dependable health service fit for the 21st century will not be able to avoid the impact of genetic and molecular science on diagnosis and treatment. The service will have to embrace everything that genetic science is able to offer by way of improving the health of our citizens. Its political masters will not be able to detach themselves from the ethical and social impact that the science will bring. Genetics cannot be ignored, and a modern and responsible government must prepare and plan for the challenges ahead.

The principle that governed the gathering of the material that is the basis of this report was that the key groups with a stake in the genetic future should be heard, together with their experience, opinions and arguments. Altogether eight groups of stakeholders took part. The project developed a standardised technique to enable groups to make their contributions. The process by which the project gathered the evidence that led to its recommendations is set out in detail in Chapter 3. A great many people gave up over half a day of their time to attend eight workshops during the summer of 1999. Those who were involved in the Policy Exploration Workshop spent two days at Winchester in the autumn of that year debating the material that had been collected, and making their recommendations.

A MODERN AND DEPENDABLE HEALTH SERVICE WILL NOT BE ABLE TO AVOID THE IMPACT OF GENETIC AND MOLECULAR SCIENCE

Genetics cannot be ignored and a modern and responsible government must prepare and plan for the challenges ahead.

This report is a distillation of the material collected during the project but does not claim to be a full description of everything that was gathered over six months of workshops, nor a comprehensive review of genetic policy. A detailed account of each of the eight workshops will be found on our website [http://www.medinfo.cam.ac.uk/phgu], together with the literature review carried out by Dr Tom Ling. These materials provide the evidence for our final recommendations. The report provides an overview of the subject, based on that evidence. By its very nature it does not focus on any individual subject. The intention was that it should reflect the views of the participants, and provide a framework for further debate. Nevertheless the findings as expressed in the report itself are the responsibility of the Nuffield Trust and of the Public Health Genetics Unit. The contribution of the participants made this possible, and to the extent that our report and its recommendations might find favour, they, the participants, must take the credit.

II

The Process

The Nuffield Trust, in collaboration with the Public Health Genetics Unit, agreed that it was essential that the evidential base for its final recommendations should be grounded in the best available informed opinion about current and likely developments in the new genetics. Once the aims of the project had been defined the project team had to decide how to gather the evidence that would be necessary to produce a properly robust set of proposals for developing a policy framework in the United Kingdom. Having identified eight groups of stakeholders they then devised a methodology that would enlist the help, and document the views of all of them.

THE FINAL RECOMMENDATIONS IN THIS REPORT ARE GROUNDED IN THE BEST AVAILABLE OPINION

The final recommendations in this report are based upon evidence collected in a series of nine workshops from individuals expert in genetics, its ethical, social and legal implications, and in health services organisation.

The Eight Stakeholder Groups

- Pharmaceutical industry
- Physicians, public health physicians, and general practitioners
- Social scientists and ethicists
- Policy commentators
- Policy makers within government and the NHS
- Medical geneticists
- Physicians with expertise and experience in genetics
- Patient representatives (GIG)

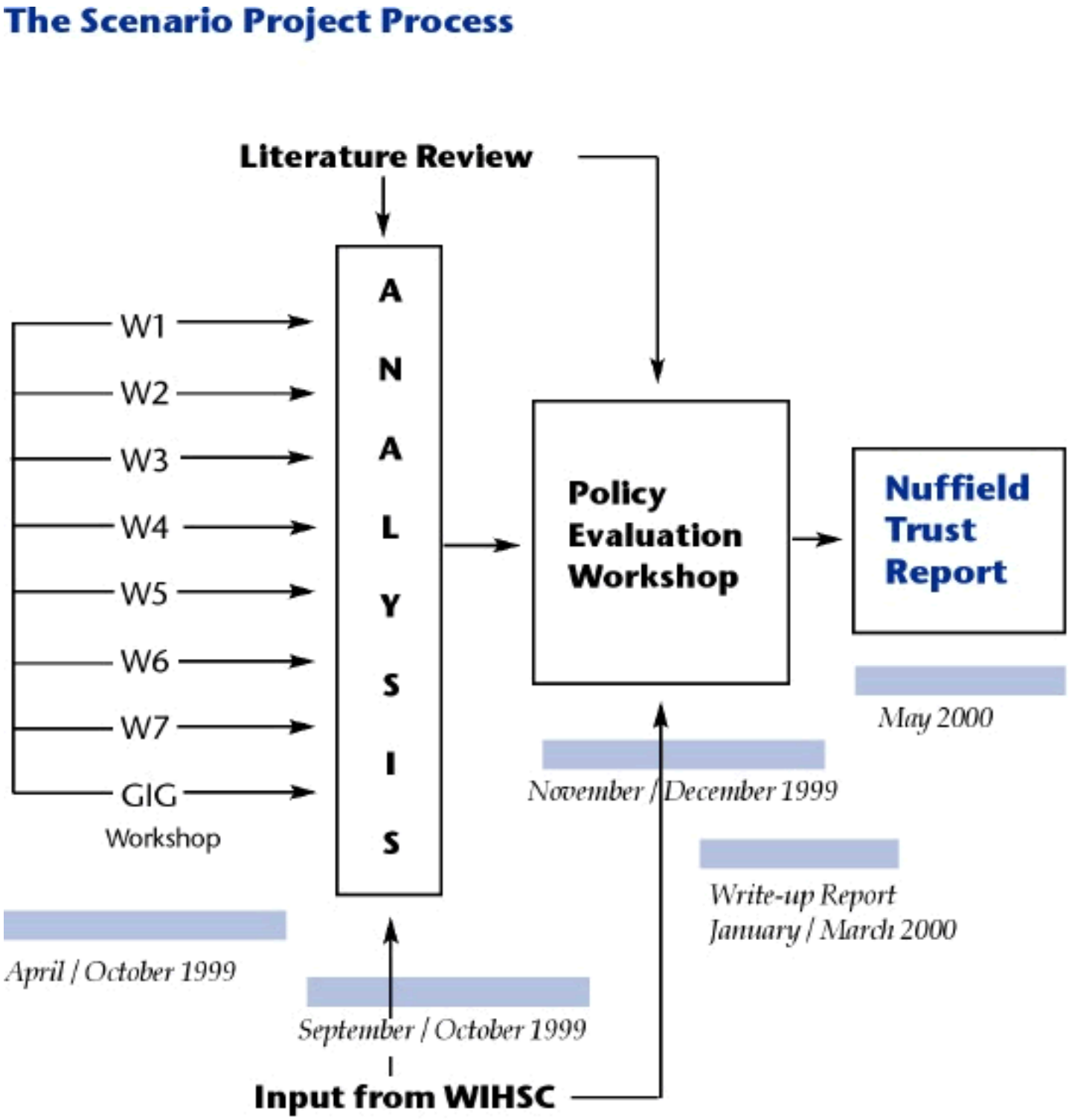
The project process was established to allow participants to each of the workshops to set out in a systematic fashion what they believed were the key issues surrounding the subject of genetics. These would be 'captured' in a standardised manner. In this way it was hoped to build into the project a method of teasing out the views of those who were responsible for planning healthcare, for its administrative and clinical provision, for providing the tests and medicines that might become available, and, to an extent, the responses of the consumer.

PUBLIC INVOLVEMENT SHOULD BE AN ESSENTIAL ELEMENT IN POLICY DEVELOPMENT

This project did not have the time or the resources to embark upon a major exercise in public consultation, but in fulfilling its principal recommendation that government must lead the creation of a policy framework for the impact of genetic science on the UK and its health services the public must be partners in the process.

In order to canvass the views of patients and their carers, the Genetic Interest Group (GIG) were invited to nominate individuals from the various special interest groups for which they are an umbrella organisation to attend each specialist workshop, as well as participating in a full session of their own. No attempt was made to involve the general public directly at

this stage. It is our conviction that public involvement is and should be an essential element in the process of policy development , but given the nature of the *Genetics Scenario Project*, and the limited amount of time at its disposal, to have involved the public in what could only be regarded as a superficial way might have laid the project open to charges of tokenism. However the publication of this report is as much for the public as it is for policy makers to deliberate and to debate, and our recommendations unequivocally state that in the next stage of the process the public must be involved.



The chosen methodology relied much on *Scenario Planning*, a way of linking the present and the future, which had already been used within the NHS. Indeed, one of the highlights of a conference organised by the National Health Service Confederation in 1997 to celebrate the fiftieth anniversary of the founding of the service was an exercise in scenario planning. This encouraged delegates to imagine their way into two quite different future worlds, one, entitled "Find my Way", in which

traditional authority was challenged by individuals who took a greater degree of responsibility for and had a greater control over their choices and their decisions, and a second, "Trust their Guidance" in which people still looked to the authority of others and of institutions to help them to make their choices and arrive at decisions.

THE DESIRED SCENARIO WILL ALLOW ALL THE BENEFITS OF GENETIC SCIENCE TO BE HARNESSED WITHOUT ANY ADVERSE ETHICAL OR SOCIAL CONSEQUENCES

The principal benefit would be better health for the population of the United Kingdom. The chief drawbacks were concerned with a failure to translate the biology that underpinned the new genetics into worthwhile medical technology, and the danger that the general public might take fright at and reject the whole subject if they were deprived of adequate information.

The main scenario that it was hoped that the stakeholder workshops would work their way towards was one in which all the benefits that would accrue from developments in medical genetics might be achieved without any of the drawbacks. The principal benefit would be better health for the population of the United Kingdom. The chief drawbacks were concerned with a failure to translate the biology that underpinned the new genetics into worthwhile medical technology, and the danger that the general public might take fright at and reject the whole subject if they were deprived of adequate information.

It was agreed that the eight stakeholder groups should be encouraged to express their views about what they felt were the key issues that were emerging currently around developments in genetics; not just the science and its related technologies, but social, legal and ethical concerns too in their very widest sense. However they were reminded that, while the project was seeking their views on the broad ethical issues that were being provoked by developments within genetic research and their possible clinical application, the whole field of bioethics was under continuing review in other quarters.

All of the eight workshops would follow the same organisational pattern. They would be held under the Chatham House Rule, which was designed to protect confidentiality and ensure that nothing that was said in the course of each half-day session might be attributed to a particular participant. It was hoped that this would encourage participants to share their expertise with the project, to speak candidly and to think out loud about 'the unthinkable'. There would be three major sessions in each workshop each of which would ask a question of participants. Inevitably the eighth workshop with members of GIG was organised slightly differently since a good many of the participants had already attended one of the previous workshops.

The Three Sessions

I. I.What do you think are likely to be the key issues regarding the use of emerging genetic technology to improve health?
II. II. What are the principal uncertainties in the immediate future that must be identified in order to develop health policies in relation to the new genetics?
III. III.What are your principal recommendations to government ministers about the development of future health policy as it might be shaped by developments in medical genetics in respect of first, national policy priorities, and second, the organisation, funding, and provision of clinical services within the NHS?

In each session the views of participants were 'caught' by a facilitator using the 'Hexagon Method'. The essence of an idea or an argument or an opinion was summarised in a short phrase and written on a plastic hexagon. These hexagons were then attached to a magnetic board where they could be organised at will into different patterns or sequences. This process can be regarded as a kind of organised 'brainstorming' that encourages people to escape conventional linear ways of thinking about problems since the ability to arrange and to rearrange the 'hexagons' is used to provoke new ways of thinking about familiar issues. Participants to the workshops were encouraged to offer as many ideas as there was time for. The workshops produced between twenty and fifty 'hexagons' during each session.

Each workshop was designed to follow a particular pathway. The first and second sessions represented steps towards a conclusion about the recommendations the eight stakeholder groups would want to make to government if health services within the United Kingdom were to harness the developments in medical genetics for the benefit of the whole population. In the first session participants were encouraged to identify the key issues that they felt attached themselves to current developments in genetics; in the second session they considered the uncertainties that lay ahead, using as a template the two scenarios that had been prepared for the NHS Confederation conference in 1997. Each workshop was reminded that an

'uncertainty' could be seen as something that was likely to make a considerable impact but the outcome of which was in doubt. These kinds of uncertainties might lead the future into quite different directions. The purpose of this exercise was to consider what might be the trigger points for radical changes in future events, to pinpoint as many of the uncertainties ahead as possible and to identify the turning point in each where events might lead in one of two different directions that possibly might be mutually exclusive.

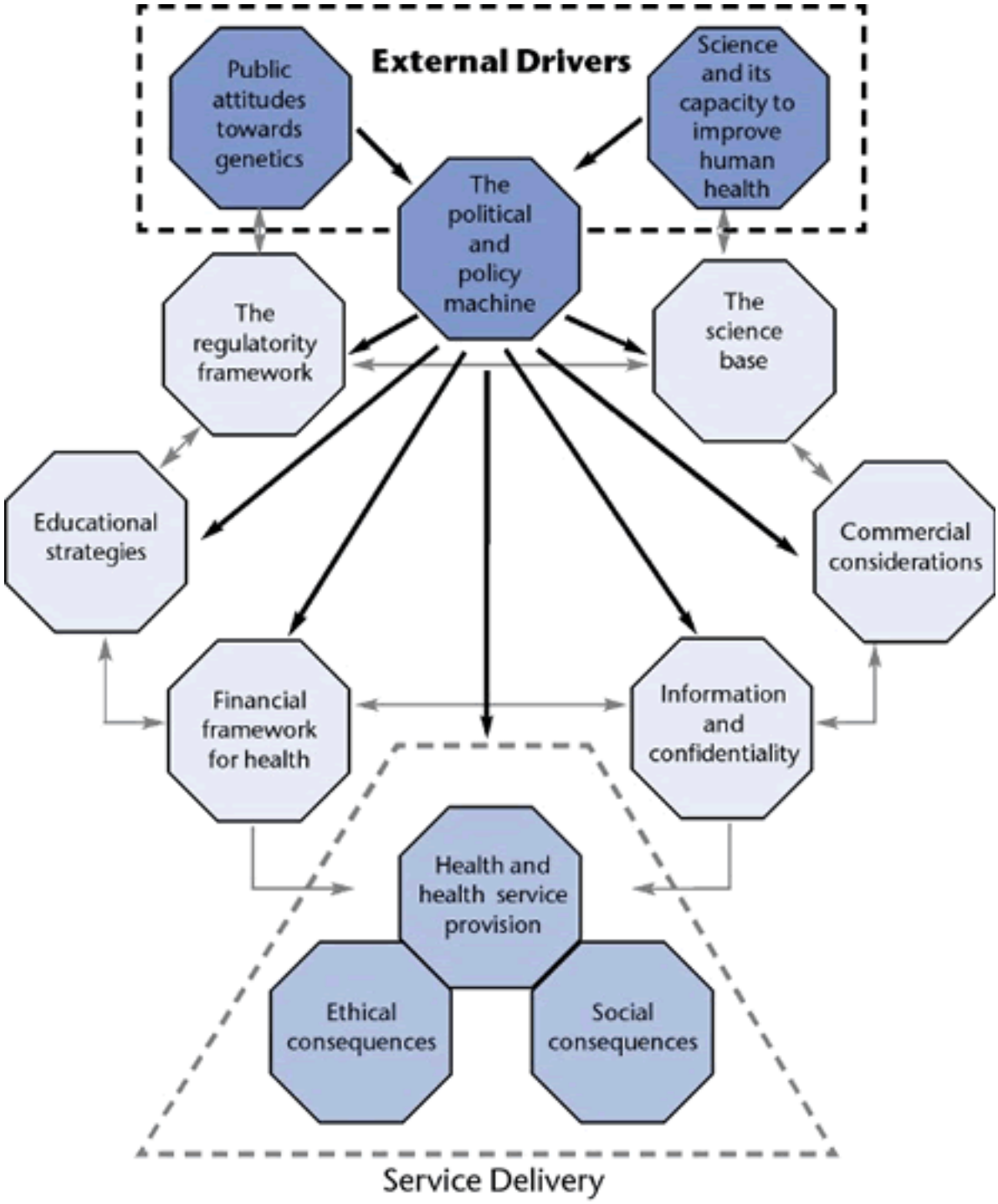
During this second exercise the uncertainties identified by the workshop were inscribed on 'hexagons' in exactly the same way as during the first exercise. What was intended was to create a route between the issues that belonged to the current state of medical genetics and a 'mapped' version of the future. That future might be hedged around with uncertainties, but at the very least it should be possible to discern its outline and its contours.

Before the third and final exercise the project team attempted to synthesise the material that had been developed so far in the workshop. What emerged from this interpretation of the data that had been collected on the hexagons was a firm idea of what it was that was driving change and the principal issues that would have be addressed as we moved into the future. In the final session of each workshop participants were asked to outline the policy priorities they now felt able to recommend in respect of future health care in the United Kingdom, and in particular the organisation, funding, and provision of clinical services within the NHS. Once the sequence of workshops had ended their proceedings were written up and drafts circulated to all who had participated in them for their approval.

The last stage of the project was a Policy Exploration Workshop held over two days near Winchester in the late autumn of 1999. Participants to this workshop were drawn mostly from the groups that had contributed to the earlier workshops. Before the workshop the project team had reconsidered all the material that had been gathered in the eight specialist workshops and identified a sequence of themes common to all of them. It was clear now that as far as current developments in genetics were concerned two external drivers *(Public Attitudes towards Genetics and Science* and *its Capacity to Improve Human Health)* were bearing down on *The Political and Policy Machine*. Then there were six major areas where future policy would have to be decided, namely *The Regulatory Framework, Educational Strategies, Financial Framework for Health, Information and Confidentiality, Commercial Considerations, The Science Base*. A seventh area was the impact of genetics on *Health and Health Service Provision and their Ethical and Social Consequences*. These specific drivers are shown in the figure overleaf.

This was the evidential base from which the project team developed a model for the conduct of the Policy Exploration Workshop. A literature review prepared by Dr Tom Ling was also available to the participants, together with the report of the *Citizen's Jury on the Genetic Testing of Common Disorders,* conducted by the Welsh Institute of Health and Social Care (WIHSC). Using the 'hexagon method' again to gather and define argument, opinion and evidence volunteered by participants, the aim of that final session was to arrive at a set of clear recommendations to be acted upon immediately, and in particular the establishment of a national policy framework that would set a context for the introduction of developing medical genetics into health services in the UK over the coming fifteen years. Their recommendations and deliberations provide the basis of this report .

Specific Drivers for Genetics


External Drivers
Public attitudes towards genetics
Science and its capacity to improve human health
The political and policy machine
The regulatority framework
The science base
Educational strategies
Commercial considerations
Financial framework for health
Information and confidentiality
Health and health service provision
Ethical consequences
Social consequences
Service Delivery

III

The Policy Context

Introduction

This chapter sets out the existing policy context for genetics and the issues associated with the subject in the United Kingdom. It deals with its implications both for the NHS and for government in its widest sense; and it touches on developments in Europe and in the United States. However it does not claim to cover these areas in detail or in any systematic fashion. Rather it is a brief review of recent history and the significant literature associated with this history. Thus it outlines the major reports that have been published in the United Kingdom, and the committees that have had an influence upon and a responsibility for many of the issues associated with genetics over the past decade. The review shows that the publications only span the last decade or so and suggests that the remarkable pace at which molecular biology has advanced have only been attended to by policy makers and governments in the last few years.

POLITICIANS AND POLICY MAKERS IN THE UK HAVE ONLY RECENTLY BEGUN TO FOCUS ON THE ROLE OF GENETICS IN MEDICAL CARE AND ON THEIR ETHICAL, LEGAL AND SOCIAL IMPLICATIONS

A review of the relevant literature shows that publication only spans the last decade or so which suggests that the remarkable pace at which molecular biology has advanced has only been attended to by policy makers and governments in recent years.

National Context

The House of Commons Science and Technology Committee Report *Human Genetics: The Science and Its Consequences*, published in 1995, was one of the earliest and perhaps the most influential of these reports. It brought to the attention of politicians and policy makers the likely impact of genetic science, as well as some of its ethical and social implications. It provided a comprehensive survey of the issues, and made specific recommendations. The government responded to these in January 1996, and, amongst other things established the Human Genetics Advisory Commission (HGAC) and the Advisory Committee on Genetic Testing (ACGT).

The House of Commons Report was not, of course, the first to address these issues. The Nuffield Council on Bioethics, in its report *Genetic Screening Ethical Issues* looked closely at the ethical and social issues of genetic testing and screening in 1993, while gene therapy had been addressed earlier by the Clothier Committee in the *Ethics of Gene Therapy* in 1992, which had led to the establishment of the Gene Therapy Advisory Committee two years later. The Committee of Enquiry into Human Fertilisation and Embryology under the Chairmanship of Mary Warnock had reported as early as 1984, and was influential in informing the Human Fertilisation and Embryology Act of 1990. This in turn led to the establishment of the Human Fertilisation and Embryology Authority (HFEA) in 1991. The HFEA has a remit to license fertility treatment involving the use of embryos created in vitro, research on the storage of human embryos, and the storage of human sperm and ova. Since then the Nuffield Council on Bioethics has examined three other issues, publishing reports on *Human Tissue: Ethical and Legal Issues* in 1995, *Animal-to-Human Transplants: The Ethics Of Xenotransplantation* in 1996, and on the *Mental Disorders and Genetics: The Ethical Context* in 1998.

The HGAC has in its turn reported on *Insurance and Genetic Testing* in 1997, *Cloning Issues in Reproduction, Science and Medicine* in 1998, and Employment and Genetic Testing in 1999. The ACGT published its reports on *Code of Practice on Human Genetic Testing Services Supplied Directly to the Public* in 1997, *Genetic Testing for Late Onset Disorders* in 1998 and *Advice for Research Ethics Committees* in 1999. More recently a draft document on *Preimplantation Genetic Diagnosis* was released for consultation in 1999, and another, on *Prenatal Genetic Diagnosis*, in 2000. In August 1999 the Chief Medical Officer established an Expert Advisory Group on Therapeutic Cloning in response to the HGAC Report on Cloning. Its

purpose was to investigate the extent of research on therapeutic cloning. Did this research raise new ethical issues; was further regulation needed to govern the use of embryonic stem cell lines and the use of human embryos in research?

In May 1999 the UK Government published a review of the *Advisory and Regulatory Framework for Biotechnology*. This led to the creation of a new strategic authority, the Human Genetics Commission, subsuming the Advisory Committee on Genetic Testing, the Human Genetics Advisory Commission and the Advisory Group on Scientific Advances in Genetics, established in 1999 by the Office of Science and Technology. The Genetic Therapy Advisory Committee will continue to provide advice on the ethical acceptability of proposals for gene therapy research on humans and to offer its opinions on developments in gene therapy research to ministers within the UK. The Genetics and Insurance Committee (see below) will similarly continue as a non-statutory body to guide insurance providers. Both of these bodies will, however, be expected to "have regard to" the Human Genetics Commission.

Most recently the House of Lords Select Committee on Science and Technology published its report *Science and Society*. While acknowledging that public interest in science was high, the report admitted that public understanding of, and indeed trust in science was not commensurate with this general interest. Among its recommendations were that existing initiatives to increase public understanding of science should be supported by government; that scientists and research students should be trained in communication skills; and that the Office of Science and Technology in collaboration with the Department of Culture, Media and Sport should take a more proactive role in establishing links between science centres, science museums and research councils while also encouraging appropriate institutions to create and maintain web sites. Scientists should be encouraged to develop their communication skills in respect of the media since most people get most of their information about science from TV and the newspapers. It was important for those institutions that were charged with responsibilities for science, particularly the research councils, to develop an open dialogue with the public. It was also essential to strengthen existing science teaching in schools. Significantly the report also argued that the Interdepartmental Liaison Group on Risk Assessment "should look into current research on how risk information is received by the public." This has a special relevance to the application of genetic science to health services.

Service Provision

We turn now to the matter of service provision and the impact of genetics on the NHS. In 1993 the Department of Health issued *Population Need and Genetic Services: An Outline Guide*. This summarised the nature of genetic disorders, the service needs of those affected by them, and the distinctive features and characteristics of a specialised genetics service. In 1995 two reviews were commissioned from the Genetics Research Advisory Group by the NHS Central R & D Committee on the future potential of genetic science for medical practice. The reports of these reviews outlined respectively advances in the field of inherited disorders and recent work on the genetics of common disorders. A report of a working group to the Chief Medical Officer in 1998 similarly considered Genetics and Cancer Services. None of these three documents resulted in any immediate change in policy for the provision of clinical services as a consequence of advances in genetics.

The Royal College of Physicians attempted to highlight the issues as they affected specialist genetic services in a series of publications since 1991. The topics that were dealt with included *Purchasers' Guidelines to Genetic Services in the NHS* in 1991, *Clinical Services into the 21st Century* in 1996, *Clinical Genetic Services: Activity, Outcome, Effectiveness and Quality* in 1998, and *Commissioning Clinical Genetic Services* at the end of that year. Clinical Genetic Services discussed the functions of a clinical genetics service; appropriate measures of activity for these services; their aims and outcomes; and the appropriate criteria for assessing quality and measuring outcomes. *Commissioning Clinical Genetics Services* set out the requirements of a good clinical genetics service with recommendations on securing these for commissioning bodies .

For its part the NHS Confederation published a report on *Commissioning Genetic Services* in 1998. This was based on a survey carried out by the Genomics Policy Unit of WIHSC. It aimed to summarise the current state of genetics services commissioning in the UK, outlining the attitudes of health authorities towards genetics services, the numbers of authorities which had genetics services either in place or planned and the existing commissioning arrangements (either through consortium arrangements or by individual contracts).

THE NHS AND ASSOCIATED PROFESSIONAL BODIES HAVE ONLY COMMENTED ON EXISTING GENETIC SERVICES

The Department of Health, the Royal College of Physicians, the NHS Confederation and other bodies have confined themselves to reporting on existing genetic services. Other bodies such as the BMA, the IPPR and the Welsh Health Planning Forum have peered into the genetic future, but to date no one has attempted to take an overview of how to plan for changes in the organisation, funding and provision of clinical services as a result of current and prospective developments in genetics.

The Welsh Health Planning Forum's *Genomics: The Impact of the New Genetics on the NHS* was published in 1993. It provided one of the earlier reviews of how genetic science would impact on the future of health services. The British Medical Association (BMA) had contributed one year earlier to the debate through its publication of *Our Genetic Future: The Science and Ethics of Genetic Technology*, and followed this with Human Genetics: Choice and Responsibility in 1998. The Institute for Public Policy Research similarly considered some of the issues at the end of 1998 in its report *Brave New NHS: The Impact of The New Genetics on the Health Service*. The School of Public Policy at University College, London published its report on *Human Genomics: Prospects for Health Care and Public Policy* in 1999. And in the same year the Office of Health Economics issued its report on *Genomics, Healthcare and Public Policy*. Issues relating to public opinion were reviewed in a study from the University of Kent at Canterbury, *Public Opinion and Regulation of the New Human Genetics: A Critical Review* in 1999. A early report of a citizen's jury in 1998 from WIHSC, *Report of the Citizen's Jury on the Genetic Testing of Common Disorders* also did much to inform policy makers of public views on the subject.

To draw attention to the users' perspective, the Genetic Interest Group (GIG), the umbrella body for voluntary organisations that represent patients with rare genetic disorders, produced its own *Guidelines for Genetic Services* in 1998. The purpose of these guidelines was "to help genetic and other service providers and commissioners, in partnership with service users, set and monitor standards, identify areas for improvement, devise strategies to develop and improve services, and plan for the future." This was preceded by GIG publications on the *Organisation of Genetic Services in the United Kingdom* and *Genetic Testing of Children* in 1995, on *Long Term Care and Insurance and Antenatal and Preimplantation Genetic Diagnosis, Embryo Research, and Contemporary Abortion Issues* in 1996, and followed by *Confidentiality Guidelines* in 1998, and *Orphan Drugs and Diseases* in 1999.

Insurance

The starting point for an exploration of the specific issues surrounding insurance may be found in the Association of British Insurers (ABI) report published in December 1997, *Life Insurance and Genetics: A Policy Statement*. The Association argued that if individuals had access to genetic information that the insurer lacked, then individuals could use this information to secure a better deal than would be the case if information were fully shared. This so-called 'adverse selection' was considered by the Human Genetics Advisory Commission in its 1997 report *The Implications of Genetic Testing for Insurance*. At that time, HGAC believed that in most cases any additional costs could be absorbed by the industry. The Commission took the view that, while the principle that premiums might be increased in line with actuarial risks was sound, only in rare cases would genetic information actually alter actuarial calculations. It is worth noting that where the predictive power of genetic information is at its strongest (single gene disorders) insurance companies already have this information through the family histories of the people they insure. With other cases, the gene-gene and the gene-environment interactions are so complex that the prospects for adverse selection would be limited. The Commission also recommended that there should be a two-year moratorium on the required disclosure of genetic test results and that an independent expression of consumers' interests should be introduced.

In its response the Government declined to accept the recommendation for a moratorium, but took the view that "genetic test results should only be taken into account by insurers in making underwriting decisions where the relevance of the test results to the risk assessment had been demonstrated". It also proposed that, "an effective mechanism should be established to evaluate the reliability and actuarial evidence relating to the use of specific test by insurers". This led to the establishment of the Genetics and Insurance Committee (GAIC) whose role was to "draw up criteria and guidance for insurance providers to submit information on genetic tests and their proposed use for insurance purposes for approval by the committee." The current position taken by the ABI which is set out in its Code of Practice is that applicants for insurance will not be required to undergo genetic tests and account will be taken of existing test results only as and when their reliability and relevance to the insurance product has been established.

THERE IS LITTLE LEGISLATION SPECIFICALLY DIRECTED TOWARDS MEDICAL GENETICS AND ASSOCIATED ISSUES.

Existing legislation or regulation has come about in response to single issues or political initiatives indirectly associated with genetics.

Statutory Provision and Legislation in the United Kingdom

There is little legislation, either primary or subordinate, directed specifically towards medical genetics and associated issues. The Human Fertilisation and Embryology Authority regulates and licenses in-vitro fertilisation and related activities, as well as experimentation in human reproduction under the Human Fertilisation Act 1990. The UK Xenotransplantation Interim Regulatory Authority (UKXIRA) advises on the regulation of xenotransplantation. The Medicines Act gives the Committee on the Safety of Medicines responsibility for ensuring the safety of drugs. The Medicines Control Agency has responsibility for the licensing of pharmaceutical products and the Medical Devices Agency for medical devices, including diagnostic tests. Genetic tests will also fall within the ambit of a new European Directive on Medical Devices with transitional proposals that come into force in June 2000.

Much significant legal activity around biotechnology is related to the issue of intellectual property rights and the granting of patents. These activities are regulated by the Patents Act 1977, the European Patents Convention, and the European Directive on the Legal Protection of Biotechnological Inventions (98/44/EU).

The Human Rights Act 1998 will incorporate the European Convention on Human Rights into English law when it comes into force in October 2000. It will have a significant impact on health policy. Article 2 of the Convention provides for a right to life; Article 3 for freedom from degrading treatment; Article 8 for the right to private and family life; and Article 14 for freedom from discrimination in the enjoyment of these rights. Pre-natal diagnosis, pre-implantation genetic diagnosis, embryo manipulation and therapeutic abortion are all likely to come under renewed scrutiny in the light of the Act.

International Guidelines

INTERNATIONAL CONVENTIONS AND EUROPEAN REGULATIONS WILL HAVE A MAJOR INFLUENCE ON THE DEVELOPMENT OF GENETIC SERVICES IN THE UK

Conventions of international organisations such as WHO and UNESCO and various European directives will be relevant to the development of genetic services.

The World Health Organisation has taken an interest in genetic issues since 1995. In that year it organised in Geneva an informal consultation on *Ethical Issues in Medical Genetics and the Provision of Genetics Services* in the light of the Human Genome Project. This led to the publication in 1998 of *Proposed International Guidelines on Ethical Issues in Medical Genetics* and of *Statement of a WHO Expert Advisory Group on Ethical Issues in Medical Genetics*. Both of these documents contained recommendations on the proper use of genetic data, the voluntary use of genetic screening and testing, prenatal testing, equity of access to genetic services, confidentiality of genetic data and the importance of education in genetics for the public and health care professionals. The WHO is at present preparing *Guidelines on Ethical Issues Relating to the Prevention and Management of Genetic Disorders*. UNESCO issued their *Universal Declaration on the Human Genome and Human Rights* in 1997. The Human Genome Organisation (HUGO) also provides a source of guidance.

Europe

The past thirty years or so have seen the growth of genetics services in most European countries. The ethical and practical issues surrounding this development may have been driven by global trends in science and technology but in each country there has been a complex co-evolution between on the one hand scientific and technical developments and on the other the ethical and institutional framework in which these developments take place.

At the trans-national level in Europe, the European Community's competence to consider genetic and biotechnology issues derives originally from treaty provisions covering the health and safety of workers, the safety of the environment, and the requirement to progressively remove barriers to trade. Article 129 of the Treaty on European Union (Maastricht) gave a specific boost to health protection issues and led to a framework for action in the field of public health in November 1993. These provisions were strengthened by the Amsterdam Treaty in 1997 which also redefined the Community's public health role. Of eight public health programmes proposed by the 1993 framework, five were adopted. Proposals put forward in 1997 have now resulted in the adoption of the remaining three, including one on rare diseases, defined as a prevalence of less than 5 per 10,000 population. This will have clear implications for genetic services.

The EU has also sought to influence the policy context in other ways. Directive 95/46/EC of October 1995 concerned "the protection of individuals with regard to the processing of personal data and on the free movement of such data", and also seeks to regulate the circumstances under which such information may be used for purposes other than those originally intended.

The UK responded with a revised Data Protection Act in 1998. The European Directive on Biotechnology (98/44/EU) states that the human body and its elements are not patentable, but that those elements that can be 'isolated from the body' can be patented. A Group on Ethics in Science and New Technologies has been established within the European Commission.

The Council of Europe's *Convention for the Protection of Human Rights and Dignity of the Human Being with regard to the Application of Biology and Medicine* was adopted by the Committee of Ministers in 1997. Its aims are "to protect the dignity and identity of all human beings and guarantee everyone, without discrimination, respect for their integrity and other rights and fundamental freedoms with regard to the application of biology and medicine." Article 11 refers specifically to the use of genetic information in unfairly discriminating against individuals, while Article 12 seeks to restrict genetic testing to health purposes or scientific research, disallowing its use for the purposes of insurance or employment. Article 13 forbids germ-line therapy. An additional protocol on the prohibition of human cloning was added in January 1998.

An early survey of genetic services in Europe by Professor Rodney Harris was unable to find sound evidence with which to document their availability within the EU. Consequently the European Commission provided funding for a Concerted Action on Genetics Services in Europe (CAGSE) to establish a network of clinical geneticists working in the EU. CAGSE then extended its network to include neighbouring countries (Yugoslavia, Croatia, Slovenia, Israel, Cyprus, Norway, Switzerland and Turkey) and in December 1997 it produced *Genetic Services in Europe*, a comparative study of activities in 31 countries which was published as a special book edition of the *European Journal of Human Genetics*. Despite some methodological difficulties, definitions of 'clinical geneticist' for example, this provided an important source of evidence on the state of genetic services in other European countries. The CAGSE report compared and contrasted genetic services from seven different perspectives: availability, access, life sustaining, non-harmful, effectiveness, state of the art, and consumer satisfaction. The report demonstrated wide variations in these perspectives, although there were methodological difficulties in making comparisons.

The United States

In the United States the philosophy embodied in the UN Convention on Human Rights has shaped much of the debate surrounding genetics. Consequently much proposed legislation has emphasised genetic information as the 'unique property' of the person to whom it relates. This principle was established through an Act of the State of Oregon in 1995. The issue of genetic privacy has since been a major issue for individual states. It has not found favour among federal legislators, despite the work of Annas and others who, in a proposed Genetic Privacy Act, asserted that the personal nature of the information contained in DNA was as personal and as private as a diary, describing "an important part of a unique and personal future".

In the absence of wide-ranging federal legislation, a number of recommendations and guidance statements have been made. The Ethical, Legal and Social Implications Working Group (ELSI) of the National Human Genome Research Institute is well known for this work along the College of American Pathologists, and the National Bioethics Advisory Board. One of the more systematic studies of the potential implications of the new human genetics comes from an ad hoc Task Force on Genetics and Disease Prevention, co-ordinated by the Centres for Disease Control and Prevention, which in October 1977 produced a *Strategic Plan for Translating Advances in Human Genetics into Public Health Action*. This document laid out a philosophical foundation, a conceptual framework and a number of goals and recommendations for service organisation and public health practice. It identified three critical issues: first, the need for partnership and co-ordination, second, the importance of ethical, legal and social implications and third, the need for education and training of health professionals.

In September 1997, another Task Force on Genetic Testing, created by the NIH Working Group on Ethical, Legal, and Social Implications of Human Genome Research, published its Final Report on *Promoting Safe and Effective Genetic Testing in the United States*. This work was followed by the establishment of the Secretary's Advisory Committee on Genetic Testing (SACGT) in June 1998 by Donna Shalala, Secretary of Health and Human Services. The Committee expects to set standards for genetic testing in the United States and, following a national programme of public consultation, submitted its interim recommendations to the Surgeon General on 15 March 2000.

These recognised the importance of the continued development of genetic tests, but stressed the need for regulation. They suggested that the Federal Drugs Agency should be involved in the review of new genetic tests and that the Clinical Laboratory Improvement Amendments (CLIA) regulations should be augmented in order that the quality and efficiency of genetic laboratories might be assessed against a nationally agreed standard. They also recognised that the public must be involved in discussing the many issues that surround genetic testing, not least in the developing debate about the clinical validity and utility of such tests. SACGT will meet again in June 2000 following which they intend to publish their final recommendations.

This brief survey of the current literature about the application of the new genetics to health service policy will need to be

considered by health planners and policy makers in the UK. It is intended to be neither comprehensive nor systematic, but seeks only to provide a context in which to set the deliberations of the *Nuffield Trust Genetics Scenario Project* as described in the following chapters.

IV

The External Drivers

Introduction

The subject of genetics has now fathered a considerable literature which more recently has provoked a policy response in the UK, Europe and the United States. The media have also played their part in bringing to public attention advances in genetic science and their impact on society. From the outset the *Nuffield Trust Genetics Scenario Project* argued that if all of these discussions and debates were to lead to an appropriate and effective way of harnessing the potential of the new genetics for better health care, then they would need to be considered in an organised manner. It was essential that current and future issues should be woven together into a coherent policy framework that would set out what needed to be decided and what needed to be done, and when those decisions should be taken and acted upon in order to maximise the potential benefit for the UK. Such a policy framework would also help to focus the current discussions around the whole subject of the new genetics.

The planning of realistic policies for the future would need to be grounded in a secure understanding of the current state of genetics research and how this might develop in the future. Equally important was how the general public was reacting and would continue to react to these developments. Perhaps one lesson to be drawn from the recent controversy about genetically modified foods was that there had been a mismatch between scientific developments in this field, public understanding of their implications and legitimate public concerns about the impact of the technology on human health and the environment. If such a mismatch was to be avoided around the subject of medical genetics then it was essential first to identify what it was that was driving change here. What were the 'drivers', those external forces that were difficult for policy makers to influence in any significant way?

A NUMBER OF GENERAL EXTERNAL FORCES WILL DRIVE CHANGES IN THE FIELD OF GENETICS

The general drivers of national and global change will affect developments in the field of genetics as in all other aspects of health care. In planning for a genetic future, the UK will need to take these into account and factor them into its policy framework.

General Drivers

It was evident that the range of external drivers that had been identified by the Nuffield Trust's Policy Futures for Health Care, and by others, as significant contributors to the shape of our future world would impact similarly on developments in genetic science and on our attitudes towards them. Advances in information technology and telecommunications, the changing role of the professional, globalisation, the impact of decisions made in Europe, environmental sustainability, human rights and demographic changes were all likely to affect our lives directly in the immediate future.

General Drivers

- Information technology
- Telecommunications
- Changing role of the professional
- Globalisation
- Europe
- Environmental sustainability
- Human rights
- Demographic changes

The developments associated with information technology and telecommunications have already provided many of the tools for the collation and statistical analysis of complex data sets, for the correlation of changes in the genome with disease and human variation and for the rapid transmission of these data between scientists across the world. The internet has established itself as a prime medium for the publication of genetic sequences within the Human Genome Project, making the results of its research instantly available on the world-wide-web. With ever-increasing access to such electronically published information, scholars, researchers and the general public have been able to keep themselves abreast of developments in the field of genetics. Its success is just one indication of an ever-growing appetite for knowledge and information, and there is every reason to expect that this trend will continue as access becomes cheaper and easier.

Armed with this knowledge many consumers have begun to develop an understanding of their own medical problems and an ability to question the authority of traditional professional opinion. Notwithstanding the difficulties of imposing order on and making sense of the sheer volume of available data, and the circumvention of the scrutiny that has traditionally validated and authenticated such work, we now observe the growth of a more confident and informed public and a change in the traditional relationship between physician and patient. In the popular word patients can now feel 'empowered' by this knowledge and choose to meet their doctor on equal terms.

These developments have also been responsible in great measure for the phenomenon of 'globalisation'. If it was economic imperatives the search for fresh markets and lower production costs that drove the old, and drives the new capitalism, it is the speed at which capital can now be moved around the world at the touch of a button, coupled with ever more contemporary economic intelligence, that effectively has abolished the old national economic frontiers. In contemplating developments in genetics it is impossible to ignore the fact that the pharmaceutical industry now operates commercially on a global scale.

Membership of a global community also imposes an obligation on individual countries to observe international conventions and guidelines. Organisations such as the WHO, the World Bank, UNESCO and the World Trade Organisation have all sought to offer guidance on economic, ethical and scientific matters. The European Union too has initiated an ever-growing volume of legislation, protocols, conventions and directives much of which has a significant impact on national law. Some of these relate directly to genetics and its ethical implications and are discussed elsewhere in this report. Despite the principle of subsidiarity, the EU has well-developed views about regulation in respect of medical research, patents, and the licensing of new pharmaceutical products with implications for all member states. As the Union enlarges to include countries from central and eastern Europe and as de facto political and economic integration grows apace it is likely that increasingly the EU will determine policy developments in the fields of science and medicine.

It is in the context of globalisation and biodiversity that we are being invited to consider regulating human activities that have an impact on the environment: not only curtailing industrial and domestic pollution, particularly from the burning of fossil fuels derivatives, that lie behind projected changes to the climate such as global warming, but also the need to husband natural resources and to switch consumption from finite to renewable resources. And arguments about sustainable resources will increasingly play a part in the development of new types of pharmaceutical products. Culturally many of these concerns about the environment reflect a new, and often moral, interpretation of the concept of the 'natural'. This has already begun to affect medical practice in the developed world where the proportion of family income spend on alternative medicines is increasing, as is the demand for so-called alternative therapies: both are regarded by many consumers as somehow more 'natural' than traditional Western medicine.

A concern for what is regarded as 'natural' has encouraged many people in the developed world to leap to the defence of the existing biodiversity of the planet which they argue is threatened by unchecked global economic development. While no one should doubt that this is a genuine reaction to a very real threat, it is most important to untangle a popular misconception that links advances in human genetics with this threatened biodiversity. Insofar as it is possible to second-guess the future it is unlikely that the biodiversity of the planet will be at risk from the application of genetic science to improving health, provided that we take care to impose appropriate safeguards within a regulatory framework.

This new perception about our relationship with the natural environment coincides with a reinvigorated interest in human rights, a belief in individual autonomy and the right to privacy, both of which will play a part in the developing relationship between physicians and their patients and the vexed question of who owns medical data.

In common with other countries in the developed world the UK is anticipating an increasingly elderly population who thanks to improved healthcare can be expected to live even longer. Indeed better health is one of the promises attached to medical genetics. However, an increase in general life expectancy will have resource implications for health services and throw into

sharper relief a possible tension between living longer and the quality of life that attends that longevity.

The Two Specific Drivers

It was clear that these general drivers would influence the development of medical genetics together with other aspects of health and health services in the UK. But were there external drivers that were specific to the subject of genetics? The answer that emerged from the workshop process was unequivocal. There were two principal drivers, separate from the general drivers described above, which together would determine the extent to which genetic science would be harnessed to bring about benefits for human health.

These major drivers were first, the nature of public attitudes towards the new science and the degree to which society would accept its ethical and social implications; and second, the science itself, and whether the greater understanding of biological mechanisms that flowed from genetics and molecular biology would lead to practical benefits and improve human health

TWO SPECIFIC DRIVERS WERE IDENTIFIED THAT WOULD TOGETHER DETERMINE THE EXTENT TO WHICH GENETIC SCIENCE MIGHT BRING ABOUT BENEFITS TO HUMAN HEALTH.

The two driving forces will be primarily determined by external factors but will be capable of being influenced by policy decisions.

Specific Drivers

1.Public attitudes towards genetics

2.Science and its capacity to improve human health

It was not the development of the science that was at issue, but whether it could be harnessed to bring about improved health outcomes in our population. These two driving forces were largely determined by external factors, but many of these external factors could and would be influenced in one or other direction by the six principal policy areas that would have to be addressed by policy makers. These six areas will be discussed in detail in the next chapter.

Public Attitudes towards Genetics

THERE IS WIDESPREAD MISUNDERSTANDING ABOUT GENETICS

Understanding of genetic issue among both the lay public and clinical professionals is poor.

Participants generally felt that there was widespread misunderstanding about genetics The unpredictable relationship between genes and environment in the determination of genetic traits and in the creation of disease was all too often ignored in favour of a simple genetic determinism as reflected in the popular phrase "it's all in the genes." This misunderstanding had to be corrected. A deterministic account of genetic science was both inaccurate and misleading, and could well develop into a form of fatalism that was likely to provoke antipathy and thus jeopardise the benefits that genetic science could bring to human health.

THE MISLEADING NOTION OF GENETIC DETERMINISM MUST BE CORRECTED

A deterministic account of genetic science is both inaccurate and misleading, and could well develop into a form of fatalism likely to provoke antipathy and jeopardise its benefits for human health.

It was clear too that public understanding about risk as it related to medicine and to other aspects of daily life was sketchy. For many the issues were both complex and difficult to fathom. Few seemed to have an overall view of the relative risks of

different activities and situations or how these affected them. Their behaviour did not appear to correlate with the probabilities calculated by epidemiologists and others, but instead to reflect their own belief systems and perceptions. Many clinicians were not particularly good at communicating the risks associated with illness and its treatment in ways that were readily understood by their patients. Often they chose to explain risk by reference to probability, and did not perceive that the way they framed their explanation whether, for example, the risk of dying from the operation was five percent, or that there was at least a ninety five percent chance of surviving it greatly affected the patient's attitude. Numerical explanation often meant very little to a public unfamiliar with numerical analysis and for whom the nature of the hazard and the severity of the outcome were of much greater importance.

THE PUBLIC FIND THE CONCEPT OF RISK DIFFICULT AND CONFUSING AND CLINICIANS ARE POOR AT EXPLAINING IT TO THEIR PATIENTS

Predicting the risk of future ill health on the basis of a genetic susceptibility will require that patients and their clinicians understand fully that concept. Adequate risk assessment and communication will be essential in agreeing upon appropriate preventive measures with patients.

This suggested that in future many people might have difficulty in grasping the significance of the kinds of risk that would emerge from genetic testing for susceptibility to common diseases that could develop later in their lives. And if they did grasp the figures, it was by no means certain that they would act on the information, either by changing their behaviour or by undertaking recommended clinical interventions. Moreover many people had lost touch with the notion that treatments as well as their behaviour were all associated with a degree of risk, that living was itself a risky undertaking!

THE MEDIA AND SCIENTISTS BOTH HAVE A VITAL PART TO PLAY IN DEVELOPING GENETIC LITERACY IN THE UK.

The media should be encouraged to resist sensationalism in their reporting of genetics. We need a new generation of well-educated science journalists who can report on genetics in an accurate and a well-informed way. We also require greater communication skills from our scientists and physicians.

The media might well play a part in dispelling public confusion about genetics. However it seemed that they too had an imperfect grasp of the subject and its impact on society. Popular newspapers were more concerned with sensation than sober reporting, while even in the more serious sections of the media there was a dearth of good scientific journalists and broadcasters, which meant that often briefings from experts were passed on to readers, viewers and listeners unfiltered by properly qualified correspondents. Scientists themselves might take some of the blame for some of the misunderstanding and misinterpretation since they were partly responsible for the hype about genetic science and its clinical potential. Enthusiasm for the subject and an eagerness to maximise its impact with one eye firmly fixed on the possibility of grant income had at times led some individuals to over-sell the technology when speaking to the media.

In the popular imagination, and also the popular media, science often seemed a kind of magic, sometimes sympathetic, but often black magic, carrying within it the threat of terrible and unimaginable consequences for all mankind. Thus public attitudes could proceed from fear, a fear of the unknown, and a feeling that developments in genetics would lead us into a chilling brave new world, a world in which, for example, the cloning of complete human beings from a single cell might be possible, and in which genetic technologists would be at liberty to pursue horrendous experiments, a world in which pigs really could grow wings and fly! These kinds of thoughts disgusted many people. Indeed many predicted developments, particularly in the field of reproductive technology, aroused feelings of distaste, described by some as the 'yuk' factor.

At the root of more considered objections to where genetic research might be leading was a range of moral and religious attitudes that regarded this science as a way of tampering with nature, and thus unnatural and wrong. It did not follow that current support for technologies designed to relieve couples from the burden of infertility, or to prevent serious genetic illness, would carry forward to embrace openly the possibility of genetic enhancement, of creating 'designer babies'.

THE NEW GENETICS IS NOT THE OLD EUGENICS

Every effort should be made to prevent the conflation of modern genetics and genetic technology with the kind of eugenics that is indelibly associated in the popular mind with Nazi Germany.

The possibility of more precise prenatal testing for genetic defects and the prospect of interventions using techniques such as pre-implantation genetic diagnosis raised another moral issue in the minds of many people. Potentially these developments would allow the detection of less severe genetic disorders, of diseases which would not be manifest until late in life, or even of physiological traits such eye or hair colour, so permitting the deliberate selection of embryos that would be implanted and the disposal of those that would not. Indeed, the mere existence of this technology gave serious cause for concern, evoking the spectre of eugenics and raising the possibility that attitudes that arose in Nazi Germany and elsewhere in the middle years of the twentieth century with such appalling consequences might reappear. Many of those who were disabled, or who represented the interests of the disabled, argued so. If genetic technologies were to permit the correction, say, of deafness or sight impairment, or the rejection of embryos with such defects, would this not stigmatise the deaf and the partially sighted? Would a new definition of what was 'normal' lead to the marginalisation of men and women who fell outside its definition?

Furthermore this notion of what was normal might be redefined as 'genetically normal', leading to the social exclusion of individuals who possessed 'abnormal' genes particularly if genetic technology only allowed clinicians to test for and identify defects without being able to correct them. While the possibility of selecting individuals on the basis of their genetic makeup would not automatically lead to such devastating consequences (despite a popular tendency to conflate the new genetics with the old eugenics), the ethical issues that were raised were serious enough to merit long and detailed consideration, and to require public discussion and resolution. To others however, such technology represented the pinnacle of reproductive choice, a triumph for humanity, and a source of hope for those afflicted by genetic disease, for whom any attempts by the state to reduce access was seen as an affront to their individual rights.

THE POSSIBILITY THAT GENETICS MAY LEAD TO NEW KINDS OF DISCRIMINATION MUST BE ADDRESSED SERIOUSLY

There is public disquiet about discrimination, particularly in the workplace and in access to insurance cover. If they are not addressed these anxieties will lead to a negative reaction against genetic science.

Uppermost in the minds of many people there was also a belief that genetic information might lead to new kinds of social and personal discrimination. The possibility that there might be discrimination in the work place and in acquiring insurance cover hinged on the question of confidentiality. Would genetic information about an individual remain a private matter between the doctor and the patient? Closer to home, would patients be able to withhold genetic information from members of their own family even when that information might have a bearing on the health status of other members of that family? These are important questions for genetic testing and its consequences that will be dealt with in a later chapter.

A number of the workshops expressed a concern about the volatility of public opinion if it were not informed by a proper understanding of genetics and the issues it raised. It might take only one example of a genetic test being abused or of a test result being misinterpreted, and for either or both scenarios to involve an individual with a high public profile for the popular media to help to turn the public against genetics and its application to health.

Taken together these anxieties would seem to constitute a powerful case for the general public turning against genetics. However there was evidence from many quarters that at present the public were broadly supportive of the new science, and indeed excited by the ways in which it would be translated into future healthcare. A recent survey from Europe reported in *Public Opinion and Regulation of the New Human Genetics: A Critical Review* from the University of Kent at Canterbury concluded that 88 percent of people in the UK supported the idea of genetic testing even if that level of support was qualified when genetic testing was related to personal issues such as insurance or termination of pregnancy. And 85 percent of those questioned also supported the use of genetic science to develop new drugs.

The goodwill towards genetics that was implied by these two statistics was a reason to be optimistic. Instinctively perhaps the public did understand the health benefits that might accrue to them from the new biology; they might be willing to conquer their doubts and fears. However, the necessary condition for putting their darker thoughts to flight had to be a radical increase in levels of genetic literacy throughout the whole population and that included medical staff as well as patients.

Science and its Capacity to Improve Human Health

Participants to the workshops were divided as to whether major developments in genetics would impact upon health and health services in the near or distant future. Nonetheless it requires little imagination to see that the remarkable advances that have taken place in molecular biology over the past two decades are dictating the pace of change in public, political and medical attitudes towards genetic science. In a very real sense this science has now achieved such a global momentum, fuelled in part by a commitment on the part of the international pharmaceutical industry to exploit its discoveries, that it has overtaken both public understanding and, more importantly, national and international policymakers and politicians.

THE PACE AT WHICH GENETIC SCIENCE WILL IMPACT ON HEALTH AND HEALTH SERVICES IS UNCERTAIN

Participants to the workshops were divided as to whether developments in genetics would impact in a major way upon health and health services in the near or distant future.

The pace of scientific development is one thing, the claims that are made on its behalf quite another. Should we take these claims at face value? Are the medical benefits that will supposedly flow from harnessing the molecular biology that underpins the new genetics probable or even possible? The mapping of the human genome is in itself no guarantee that in the future common diseases will be predictable, and so preventable. In the opinion of a great many participants to the workshops it was essential to imagine a future in which the science associated with genetics might not be able to deliver on its technological promises, at least not within a fifteen-year timescale.

The paradigm of the rare single gene disorders, conventional genetic diseases such as cystic fibrosis or muscular dystrophy, which passed from one generation to another in a manner predicted by the laws of inheritance, did not apply in general to common diseases. The situation in which the disease state, or the genetic trait, followed the passage of a single abnormal gene was the exception rather than the rule. These so called "genetic diseases" were rare, but did follow a course in which those members of the family who had inherited the abnormal gene by and large developed the disease, and those who inherited the normal version did not. Medical genetics was the specialty that dealt with these rare single gene disorders. But even in these examples, correlation was not perfect. In some cases the possession of the abnormal gene did not result in disease; in others the severity of the disease and its manifestations varied significantly between family members, even though the genetic abnormality was the same.

For common disorders such as heart disease, cancer or diabetes, it was clear that there was no pattern of inheritance among family members, even though the presence of the disease in one member of the family statistically increased the risk of the disease in another. Twin studies that compared the frequency of disease between identical and non-identical twins also provided evidence that a genetic component was at work. The role of abnormal genes in the determination of disease risk was probabilistic; their presence only accounted for an individual's increased susceptibility to disease. Disease could not be predicted just from knowing that an abnormal gene was present. Disease arose because a multiplicity of genes interacted with each other, and with the external environment.

At present we had an imperfect understanding of gene-environmental interaction in the determination of disease. It was one thing to maintain that every common disease had a genetic component; it was quite another to explain why, for example, in two people who both possessed an abnormal gene that had been linked to a particular illness, one might develop the disorder while the other remained free of it. The precise contribution of environmental factors such as diet, pollutants and infectious agents to the pathogenesis of disease was still unclear. It was equally unclear how genes acted upon each other to create an opportunity for disease to develop.

Medicine had traditionally relied upon the careful observation of patients and the recognition of phenotypes, physically observable human traits or manifestations of disease, as the method by which diagnoses were made. With time, and greater scientific understanding, diagnosis became more sophisticated, moving from a descriptive to a pathological and ultimately aetiological categorisation of disease. One of the promises that attached itself to medical genetics was an entirely new ability to categorise disease and to develop in future years a classification based on genotype instead of one that depended on clinical and pathological characteristics.

SOME BELIEVE THAT GENETIC AND CELLULAR MECHANISMS ARE TOO COMPLEX TO ALLOW COMMON DISEASES TO BE PREDICTED OR PREVENTED IN THE NEAR FUTURE

The relationship between individual genes and between those genes and environmental factors in the creation of common diseases is exceedingly complex.
It may prove difficult to make the necessary correlation between genetic variation and disease states. The resources required to establish a correlation between genotype and common disease may just place too great a burden on the national economy.

There was also a promise that one might be able to establish correlations between genotype and phenotype, between observable characteristics and an individual's genetic identity. As yet there were considerable difficulties in establishing that correlation, and the view was held in some quarters that it would not become any easier to do so in the immediate future. These doubts stemmed from the complexity of the disease process and of human biology, and from knowing that an accurate correlation between genotype and phenotype would require large population samples, huge amounts of computing power, considerable expertise in bio-informatics and significant investment in an infrastructure for the collection and storage of DNA samples. Would society, in the next decade or two, be able to mobilise the resources to meet this challenge and achieve worthwhile and clinically relevant results? Yet this was one of the corner stones on which our hopes for medical genetics was built. It was clear that mapping the human genome and then individual genes was only a beginning, and that if the science associated with genetics was to be able to sustain its present level of development we would need continuing investment in research on a considerable scale, nationally as well as globally.

Depending on the individual's perspective, these concerns about the science could be described either as a pessimistic version of the future or a realistic assessment of what lay ahead. But prediction is always problematic.

The optimistic version of events argued that the ability to correlate genotype and phenotype would be translated into tangible medical benefits sooner rather than later, that a greater understanding of biological mechanisms would enable a therapeutic future in which drug treatment would be individualised for patients and in which gene therapy, cloning, and stem cell technology would provide a novel and effective way of treating certain diseases. The use of chip technology to provide a rapid scan of the human genome or of the expression of genes in human tissue would enhance our ability to diagnose disease, and enable an accurate stratification of risk in individuals. Its use by the practitioner would not require a detailed understanding of genetic mechanisms, but only an ability to recognise patterns on the chip and the ability to correlate those patterns with disease.

THE OPTIMISTIC VIEW OF GENETIC SCIENCE ARGUES THAT WE SHALL SEE TANGIBLE BENEFITS IN PREDICTION AND PREVENTION WITHIN FIVE TO TEN YEARS.

Developments in chip technology will give us the ability to scan the individual genome for susceptibility to disease ever more rapidly. A greater understanding of biological mechanisms will herald a therapeutic future in which drug treatments will be individualised for patients, and in which gene therapy and stem cell technology will provide new and effective ways of treating certain diseases.

Within a ten-year period medical care would have changed beyond recognition. It would herald a future in which genetic data on a chip providing over the counter test results and information freely available on the internet would enable citizens to bypass their general practitioners and go directly to specialists. The pharmaceuticals industry would have translated genetic science to produce an entirely new range of drugs as well as diagnostic tests to predict the effectiveness of their products or to identity those who might be susceptible to adverse side effects. We should be preparing for unprecedented changes in both the structure of health services and the way in which they were delivered. The science associated with genomics would launch a revolution in healthcare akin to that defining moment in nineteenth century medicine when the significance of infectious diseases was finally understood. What we should be preparing for was nothing less than a shift in the paradigm of medical practice from diagnosis and treatment to one that would be based on prediction and prevention.

How then might policy makers devise a policy framework that would keep pace with scientific developments, given all the uncertainties? When precisely 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 empowered patients and their clinicians to predict the likely onset of common diseases, how would that patient be persuaded that it was wise to adopt a preventive strategy when evidence suggested that most people chose to ignore the risks attached to certain lifestyles?

Whether one took an optimistic or a sceptical view of the genetic future, both positions imposed obligations on those who

would plan for the future. First, there was a responsibility to understand what they wanted science to achieve and its trade-offs against other social values, since scientific activity, like all other social activities, took place in a cultural, political and economic context. Second, genetics was 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. Our understanding of single gene disorders had advanced steadily over the past quarter of a century, but we should not assume that a comparable understanding, say, of the relationship between low penetrant genes and common diseases would advance so smoothly.

V

The Six Policy Areas

Introduction

SIX POLICY AREAS HAVE BEEN IDENTIFIED OVER WHICH GOVERNMENT AND OTHER STAKEHOLDERS WILL BE ABLE TO EXERCISE A DEGREE OF INFLUENCE

Decisions taken within these six identified policy areas will have a significant impact on how and whether the two specific drivers (Public Attitudes and Science) will lead to improvements in human health.

The project had identified two specific drivers, public attitudes towards the new genetics and the emerging science which between them would determine how the science might impact on the delivery of health care. At the conclusion of the stakeholder workshops it was also evident that of the many matters raised by the participants, the majority could be categorised into six policy spheres over which government and others involved in policy development would be able to exercise a degree of influence. The six policy areas are summarised below and form the subject matter of this chapter

The Six Policy Areas

1.The Regulatory Framework

2.Educational Strategies

3.Financial Framework for Health

4.Information and Confidentiality

5.Commercial Considerations

6.The Science Base

Other issues, which will be discussed in the two subsequent chapters, related either to health service provision and its ethical and social consequences, or to the process of policy development itself. Here participants to all the workshops were clear that the nature of the policy making process, the values that were to inform it, and the means by which the various stakeholders were to be involved, were as important as the policy issues themselves.

DECISIONS TAKEN IN ONE POLICY AREA HAVE IMPLICATIONS ACROSS THE WHOLE SPECTRUM OF POLICY

Participants realised the interdependence of the key policy areas in which future policy would have to be defined and refined. What was decided about one key area would have an impact on decisions made in each of the others.

While the participants at the final policy exploration workshop unanimously accepted the categorisation, they also recognised

the interdependence of the key areas in which future policy would have to be defined and refined. It was essential to be clear about how each interacted with the others. For example, the kind of science base that the UK might sustain would be determined in equal measure by public attitudes to genetic research, by the investment decisions of the commercial sector, by the speed at which the science translated into real benefits for the UK population and by the financial regimes of the UK health system and the extent to which it would have the resources to purchase these emerging technologies.

The Regulatory Framework

A REGULATORY FRAMEWORK TO COVER ALL THE HEALTH IMPLICATIONS OF THE NEW GENETICS WILL BE NEEDED

There should be a co-ordinated regulatory framework to cover both the statutory and non-statutory health implications of genetic science which would take the long view of likely future developments.

The regulatory framework that we would adopt within the UK was a matter of concern to all of the participants to the workshops. They were united in their view that such a framework was needed to cover all the implications of the new genetics and that regulation here would need to take the long view of likely future developments. They recognised that the field was not new, and that the UK was not devoid of policy recommendations. An advisory structure was already in place. International conventions and protocols abounded while organisations such as the Royal Colleges, the British Medical Association, the British Society of Human Genetics, the Genetic Interest Group, and various others had published guidance about genetics. However many of the participants were of the view that despite this activity the issues surrounding regulation were by no means clear. Many of the bodies identified had an advisory rather than a regulatory role. There appeared to be little in the way of statutory regulation, and equally little co-ordination between the various national and international initiatives.

SOME REGULATION ALREADY EXISTS

The UK is not devoid of policy in this area. An advisory structure is already in place, and some policy initiatives have led to regulation in specific fields.

REGULATION IS NEEDED TO PROTECT THE INTERESTS OF THE PUBLIC BUT SHOULD NOT BE SO TIGHT AS TO PREVENT PROMISING DEVELOPMENTS

The safety and security of the public should be the primary concern of all regulation, but an appropriate balance between controlling and enabling developments in genetics should be maintained.

The participants also recognised a dilemma for any regulatory framework, that the 'controlling' element within its procedures might dominate its 'enabling' role. That is if too light a regulatory hand might put the public at risk, then one too severe could actually retard promising developments in genetics. It was essential in devising and administering regulatory mechanisms to maintain a balance between protecting the citizen and encouraging the science, and to chart a 'middle way' between the Scylla of prohibition and the Charybdis of complacency. It would not be to anyone's advantage either to ban the responsible implementation of science or to fail to prevent its adverse consequences. Everyone agreed that the primary aim of regulation should be to protect citizens and their interests, to ensure their safety and to guarantee their security. It also had an important part to play in the maintenance of quality.

It was well understood that since so much of what was happening in the field of genetics was at a global rather than at an exclusively national level, and that so many of the players in Britain had international ties or identities, the UK could not expect to establish its own regulatory system without regard to decisions taken in other countries. It would be advantageous to act in concert with our partners in Europe and in North America, and where possible to harmonise with them. If circumstances were such as to make that difficult, it was essential that our voice be heard clearly in every international forum in which these issues were deliberated and decided upon.

REGULATION SHOULD HAVE REGARD TO INTERNATIONAL TRENDS AND DEVELOPMENTS

Much of what was happening in genetics was at a global level. The UK could not expect to establish its own regulatory system without regard to decisions taken in other countries.

Within a devolved United Kingdom in which responsibility for service provision was now divided between London, Cardiff and Edinburgh, regulation would need to take account of these regional differences. Even though the lead for genetic policy was vested in Westminster, it was likely that there would be some disparities in both the funding and organisation of genetic services. However, participants believed that the general principles that informed regulation should be common to all new political entities in these islands. It would be important too that both the NHS and services within the private sector, and their ancillary activities, which touched upon genetics were regulated according to the same principles, and that both were encouraged and restrained under the same regulatory rubrics informed by a common core of values and principles.

REGULATION THROUGHOUT THE UK AND ACROSS BOTH PUBLIC AND PRIVATE SECTORS SHOULD BE INFORMED BY THE SAME VALUES AND PRINCIPLES

Despite devolving responsibility for their own national health services to the Scottish Parliament and the Welsh Assembly, the Government of the UK should ensure that regulation in the field of genetics across both the public and private sectors is informed by a common core of values and principles.

Regulation as it applied to the pharmaceutical industry would need to be appropriate and constructive. While no one doubted that we should expect that regulation would demand an increasing burden of proof about the efficiency of the diagnostic tests and the treatments that were brought to the market, that burden should not be so onerous as to deter technological initiative. It was also necessary for regulation to keep pace with technology. How should we ensure that national regulators in the UK were in possession of adequate and up to date information about genetics?

If, as had been predicted, a number of genetically based diagnostic tests were to be openly available to the general public, sold over the counter by chemists and pharmacies, what then would be the criteria governing their use? There was also a distinct possibility that such products might be purchased from beyond national borders. How would regulation apply in this situation? And what would be the place of genetic counselling, which had hitherto been generally agreed to be an essential part of testing procedures for single gene disorders? Would regulation have a part to play in insisting that testing must be accompanied both before and afterwards by adequate counselling no matter what the circumstances surrounding the test itself?

The accumulation of information from testing, together with other genetic information about individuals, their families, or the wider social groups to which they belonged, raised ethical issues that that would need to be addressed within a regulatory framework, as well as more specifically in the policy areas that were concerned with *Information and Confidentiality* and with *Service Provision*. What manner of regulation would be required to protect a patient's genetic information; what should be the rules that ought to determine the ethical handling of that information; and who might and might not have access to it?

REGULATION MUST ADDRESS ETHICAL ISSUES AND BALANCE THE INTERESTS OF INDIVIDUALS, FAMILIES AND SOCIETY

Regulation should balance the needs of individuals, their families and society.

Some areas of research and practice were already subject to stringent regulation: human fertilisation, cloning and gene therapy for example. Might we expect pressure in the future to lift the prohibitions that had attached themselves to research in both of these fields; or might society seek to impose greater restrictions? Similarly it was likely that we would see the potential for considerable advances in reproductive technologies, of which pre-implantation genetic diagnosis provided one example. The ethical position that was adopted towards such advances would have to be transformed into practical regulation. A balance would have to be made between the needs of individual couples, their autonomy of choice, and what society deemed to be acceptable. In the more distant future there would be ethical and therefore regulatory issues to be resolved around the

possibilities of genetic enhancement, gene replacement and perhaps germ line modification if it proved practical to both correct a mutation and prevent its transmission to future generations. It was neither possible nor desirable to predict the exact way in which technology would develop. One could not anticipate its impact on public and social attitudes and it was impossible to be certain about the impact of global trends in regulation and how these might interact with national requirements. A firm set of regulations could only be developed in response to developments and issues as they arose.

Nevertheless, the participants felt that it was important to establish a set of principles to guide those who had the responsibility for promulgating regulation, and to ensure that there was a balance between the requirements of science and commerce and the duty of governments to protect the citizen. The principles would have to acknowledge, among other things, the importance of responding flexibly and speedily to new situations, and enable the UK to take full advantage of its capacity in research and development, protect the health interests of the general public and allow moratoria to be imposed on activity deemed unacceptable for whatever reason. It was this that they wished to make a priority recommendation for government action.

Educational Strategies

THERE SHOULD BE A CONCERTED PROGRAMME TO RAISE THE LEVEL OF GENETIC LITERACY IN THE UK

The issue of genetic literacy within the UK is of paramount importance both in the shaping of public attitudes and in the education of health professionals, and should be addressed at the earliest opportunity.

Participants at every workshop emphasised the importance of establishing and implementing a national educational strategy. There was widespread professional and public misunderstanding about the subject of genetics. The issue of "genetic literacy" within the UK was of paramount importance in the shaping of public attitudes, and it was essential to address this matter at the earliest opportunity. The word 'genetics' harboured many different ideas, which needed to be distinguished and unpacked before misunderstandings hardened into prejudice of the kinds described in the previous chapter. The distinction between single gene defects as causes of conventional genetic diseases and the idea that genetic factors contributed to all human variation and disease needed to be understood; so too that both genes and environment and their interaction were responsible for the pathogenesis of complex diseases such as diabetes, cancer and heart disease. These concepts were essential both for an appreciation of modern genetics and to dispel the notion of genetic determinism. There were still many who failed to comprehend the nature of gene-environmental interaction and its role in modulating the correlation between genetic variation and the presence of disease. Genetic tests would have an increased role in the future. Their place in the diagnosis of symptomatic patients, in the prediction of disease in asymptomatic individuals and in the determination of susceptibility in those with a genetic predisposition had to be clearly set out. The public would also need to understand much more precisely the nature of risk.

POLITICIANS HAVE A RESPONSIBILITY TO LEAD THE PUBLIC DEBATE ON GENETICS

Government should set an open and wide ranging agenda for discussion about current and future developments in genetics.

It was felt that politicians had a responsibility to lead the public debate, setting an open and wide-ranging agenda for discussion about current and future developments in genetics. There was an unanimous view that there should be a concerted campaign to raise public understanding and awareness. If this process were to be properly managed there was no reason to suppose that genetics should not continue to command wide support amongst the electorate. The consequences of failing to carry public opinion would be grave: the wholesale rejection of genetically modified foods provided a warning of what might happen. The media too would have a part to play in developing this climate of genetic understanding amongst the public, though reservations were expressed about how knowledgeable they were, and in some cases whether they might be able to resist the sensational in favour of a more considered approach to the reporting of genetic discoveries.

TRAINING AND EDUCATION IS NOT A ONE WAY PROCESS AND MUST INVOLVE GENUINE DIALOGUE AND DISCUSSION

The notion that training and education is a one-way process by which the 'ignorance' of the lay public can be dispelled by

dictating scientific or medical information to them is unacceptable. It is not an appropriate way of ensuring an effective public understanding of science.

The notion that training and education was a one-way process, by which the 'ignorance' of the lay public could be dispelled by dictating scientific or medical information to them was unacceptable. It was not an appropriate way of ensuring an effective public understanding of science. Of course, there was a basic knowledge that we would all need to acquire, but if we hoped to encourage genetic literacy it would have to be achieved through dialogue, discussion and an open exchange of views. The public and professionals, scientists and physicians, would need to work in partnership, with both partners taking note of what the other wished to contribute to the learning process.

There was broad agreement that in a more formal educational context modern biology and an understanding of risk should be incorporated into the national curriculum as soon as was practical. It was considered appropriate for this to start in primary schools and to continue through secondary education. The generation just beginning its formal education would be leaving school or embarking on higher education in twelve to fifteen years, the very time when the genetic revolution was predicted to make its major impact on health services. We had a rare opportunity to inculcate an informed understanding of the concepts of genetics and associated issues in the minds of a whole new generation of citizens. An appreciation of risk was surely an essential part of citizenship.

SOCIETY WILL NEED TO BE BETTER EDUCATED ABOUT RISK AND ITS RELATIONSHIP TO HUMAN HEALTH

An understanding of risk should be incorporated into the national curriculum as soon as is practical. An appreciation of risk is an essential part of citizenship.

At universities it would also be appropriate to review the disciplines for which curricula should be modified. It would certainly be necessary for medical students, nurses and other health professionals in training to be given significantly more training in genetics and in concepts of risk than hitherto. Genetics should no longer be regarded as one of many sub-specialties, which might or might not be assimilated by the student, but an examinable core component in the training of all clinicians. No one should doubt the importance of getting a first generation of genetically trained clinical staff in place a quickly as possible, or the advantages of formal education as opposed to in-service training at a later date. Equally it would seem to be desirable that a knowledge of genetics and the concept of risk be included within the curricula for subjects as diverse as, say, law, philosophy, sociology and theology.

BIOLOGY SHOULD BE A REQUIRED SUBJECT AREA IN GENERAL AS WELL AS SPECIALIST EDUCATIONAL CURRICULA

The new biology should be taught in primary and secondary schools and across a range on university subject areas.

Elsewhere the ever-growing popularity of distance learning packages should be used to promote wider genetic literacy. Equally the current political commitment to ensuring that every child of school age and the majority of homes in the UK had access to a computer and to the internet as an essential source of information could be harnessed to these educational aims.

ALL HEALTH PROFESSIONALS INCLUDING HEALTH SERVICE MANAGERS WILL REQUIRE EDUCATION AND TRAINING IN GENETICS.

Everyone associated with the provision of health care in the UK will need to be provided with a general education about genetics and re-skilled for the practice of a new kind of medicine.

It was equally important to prepare all those who worked within the health services for the anticipated changes to clinical

practice and the consequent demands these would make on service organisation and funding. Clinicians in both hospital and primary care settings, and policy-makers and health service managers at local, regional and national levels, would require in-service training to increase their awareness of the implications of the new genetics as it impinged on their particular areas of practice and responsibility. Politicians, civil servants, hospital managers, commissioners of services, public health physicians would all need to understand the impact of genetics in order to discharge their responsibility for making funding available for the necessary professional development of health service staff, and for implementing the changes required to support the a new paradigm in the delivery of clinical services. Additionally, they would need to acquire an understanding of the ethical, legal and social implications of these advances, since these, quite as much as the technicalities of the science would determine how services would develop.

Clinicians would similarly have to prepare for their changing role within the clinical consultation, and to equip themselves with skills in risk assessment and communication. It would be essential for them to be able to explain clearly to patients their individual risks of common diseases as predicted by genetic tests, and to have a clear understanding of the social and psychological impact of such information on them and on their families. Physicians, surgeons, general practitioners, nurses, health visitors, midwives and other clinical professionals would all require appropriate training.

As public views were increasingly included in the formulation of health service policy it would be essential that any member of the public who had a formal relationship with health service mechanisms should be properly informed about genetics. The lay members of health authorities, primary care groups and community health councils would all need to be coached on the subject of medical genetics and its likely effect on the provision and delivery of services. Likewise, members of local research ethics committees had a particular responsibility for ensuring that they were well schooled in issues of genetic science and their ethical implications.

MEDICAL GENETICISTS AND GENETIC ASSOCIATES WILL HAVE A CRUCIAL ROLE IN THE EDUCATION AND TRAINING OF OTHER HEALTH PROFESSIONALS

The manpower and resource implications of a comprehensive educational strategy will be considerable. Medial geneticists and genetic associates will have a crucial role to play in the development and implementation of this agenda.

The manpower and resource implications for such a wide ranging educational programme would be enormous. Consideration would have to be given to how this huge amount of training and education would be undertaken, and by whom. Medical geneticists and genetic associates would have a crucial role to play in this endeavour. Their clinical workload was already increasing at an unprecedented rate, and the burden of taking on an enhanced responsibility in training could only be accomplished if there was an increase in their own numbers. It was not adequate just to pay lip service to this requirement. A formal strategy with clear objectives had to be developed and its implementation to be adequately resourced.

Information and Confidentiallity

Personal information about an individual's health status is private. An obligation of confidentiality arises within the relationship between a health professional and a patient. This obligation imposes a duty on that professional not to reveal personal information about a patient without the patient's explicit consent. The obligation is protected by statute, the common law, numerous ethical codes and professional guidance issued by the General Medical Council. Together they provide necessary safeguards for the protection of personal health data. The advent of computerised records capable of storing enormous quantities of personal information has intensified the need to protect the confidentiality of the individual citizen.

PERSONAL HEALTH INFORMATION SHOULD CONTINUE TO BE PROTECTED BY THE FULL FORCE OF LAW

Genetic information is personal and should be protected in the same way as any other type of health information.

However, the protection of confidentiality is not absolute. The legal framework within which confidentiality is guarded and guidelines from the GMC both allow for circumstances in which such information may be disclosed without explicit consent. The notification of communicable diseases, the sharing of medical information with other health professionals in the course of caring for a patient, when ordered to do so by a court of law and where it may be justified as being in the public interest

provide examples of due cause for disclosure. Of these the issues most relevant to genetics are those which relate to consent and to the public interest.

Participants to the workshops were unanimous in their view that all personal health information should continue to be protected by the full force of law. The discussions that followed surrounded the nature of genetic information and if there were any grounds for treating it differently from other kinds of personal health information. Was the former somehow intrinsically different from the latter? Were there grounds for affording genetic information higher levels of protection? Should we subscribe to the concept of 'genetic exceptionalism'?

IS GENETIC INFORMATION DIFFERENT FROM OTHER KINDS OF HEALTH INFORMATION?

The phrase 'genetic exceptionalism' is used to describe a view that genetic information is quite different from other kinds of health information. There is no agreement about this matter. We believe that it is important for all medical information to appropriately protected.

Some argued that genetic information about an individual was different because it carried implications for other family members; and therefore since so much more was at stake it required a greater degree of protection. At the same the shared nature of this information provoked questions about whether the rights of the individual were absolute in this case. The traditional supremacy of individual rights in a legal as well as a purely ethical sense had to be tempered by an understanding of the individual's wider family and social responsibilities, and the individual's interest balanced against family interests and the public interest. No man or woman could expect to be an island in all of his or her new medical circumstances. The Genetic Interest Group had argued most cogently that genetic information belonged to the family, and that the unit of confidentiality for genetic information was the family and not the individual. The results of genetic testing, which enabled a diagnosis of a genetic disease to be made, had implications not just for the patient but for other family members.

HAVE INDIVIDUAL PATIENTS A RIGHT TO KEEP THEIR GENETIC STATUS PRIVATE IF INFORMATION ABOUT THAT STATUS HAS HEALTH IMPLICATIONS FOR MEMBERS OF THEIR FAMILY?

At present the law and medical ethics guarantee confidentiality in dealings between patients and doctors. But genetic information about an individual may have important implications for other members of his or her family. In the future it may be necessary to negotiate a satisfactory balance between the interests of individuals and those of their families.

Genetic information was also perceived to be different because it contributed to what one commentator had christened a 'future diary'. DNA was a kind of coded probabilistic future diary since it described an important part of an individual's unique future, and as such might affect that individual's current view of their life or how it might develop. The perception was that genetic information was unique and instinctive in its ability to offer patients a glimpse of their future health status. Whether this was so or not would depend upon the condition. While the special nature of genetic diseases and their relationship to highly penetrant single genes would permit such prediction, generally this was not the case.

Others were sceptical about the idea that genetic information should be better protected or handled differently. Testing successfully for genetic susceptibility to the onset of most common conditions was scarcely prophecy given our uncertainties about the potential of genes of low penetrance to predict disease. Moreover, the use of non-genetic tests could also lead physicians to predict likely health outcomes for their patients. High cholesterol levels were known predictors of cardiovascular disease, and high blood pressure of cerebrovascular disease risk. These tests had not merited special levels of protection. If special levels of protection were required then it should be in respect of monogenic disorders of high penetrance and not genetic information in general, whether that information came from the results of testing or from other sources.

So was it the possibility that genetic testing might reveal significant information about a patient's parents, siblings and other relatives that made this information different from other forms of medical data? But genetic information was not unique in this respect either, since without recourse to genetic testing familial aggregation was discernible not only in the monogenic disorders but also in a range of common disorders including heart disease, cancers and diabetes.

A further difference between genetic and other medical information cited in a number of workshops reflected concerns that the

susceptibility to a particular disorder revealed by a test might be used to stigmatise individuals and thus lead to new forms of discrimination. This, however, seemed to some a circular argument since stigmatisation and discrimination would only occur if genetic knowledge were to be regarded as special. Furthermore, many conditions that had yet to reveal an exclusively genetic component were potentially a starting point for discrimination, depression or heart disease in relation to employment for example. The logical conclusion of this position was that it was reasonable to discriminate on the basis of non-genetic factors, but unfair to discriminate on genetic grounds.

It is our view that those who believe in genetic exceptionalism are falling into the trap of dividing disease into two categories, genetic diseases and non-genetic diseases, when it is apparent that most diseases are the product of a combination of genetic and non-genetic factors; and that if some non-genetic factors are a matter of personal choice, for example smoking and hang gliding, others are often beyond our control polluted air and exposure to radon. As the chair of the Taskforce of Genetic Information and Insurance, a subgroup of the official working party established in the United States to consider the Ethical, Legal and Social Implications of the Human Genome Project, has observed,"Genetic information is special because we are inclined to treat it as mysterious, as having exceptional potency or significance [and] not because it is different in some fundamental way from all sorts of other information about us". However even if genetic information is no different in kind from any other medical knowledge it should nevertheless still be treated as both confidential and private and with the utmost respect.

WHAT RIGHTS SHOULD DONORS HAVE OVER THE RESEARCH PURPOSES TO WHICH THEIR STORED GENETIC DATA OR TISSUES ARE PUT?

Both the UK and the EU have taken legislative steps to protect citizens' data on the basis that the individual has an absolute right of privacy. Some argue the public interest requires that suitably coded or anonymised genetic data must be made available for bona fide research. A balance will need to be struck between these two extreme positions.

How a belief in absolute confidentiality might affect the creation of genetic databases for population and epidemiological research was also discussed in a number of workshops. Information gathered and stored from both individual tests and from population cohorts, whether it was collected for a specific piece of research or culled from existing records could be of great benefit in establishing the correlation between genotype and disease. What rights, therefore, should a patient have who had given blood or a tissue sample for testing over that sample? Were any rights retained over the purposes to which it could be put? How were the benefits that would accrue to society from the fruits of population-based research based upon further examination of these kinds of sample to be balanced against the confidentiality of the individual's personal data and his or her tissue specimens?

The participants all believed that explicit consent had to underpin discussions about research and the use of personal information. This was not to say that there were no circumstances in which genetic information could be used without the subject's consent, but arguments to support such exceptions had to be justified and examined in the most rigorous manner. There were also special issues about using infants and incompetent individuals as research subjects that were not discussed in detail but were nevertheless highlighted as being of the greatest importance.

The Medical Research Council had developed a set of guidelines on *Human Tissue and Biological Samples for Use in Research*. To date the Council had identified three different categories of biological samples.

- Identifiable biological samples that could be immediately linked to a specific donor
- Coded but traceable biological samples that might be protected by a coded or encrypted tag system, but which could be linked to a specific donor following the breaking of a code
- Anonymised biological samples stripped of all identifiers

Anonymised samples offered the greatest protection to the individual, but were the least useful for medical researchers for whom traditional medical records allied to genetic data were the best way of proceeding. So, given that coded samples were more valuable, which institutions, public or private, were likely to be acceptable to the public as custodians of these data, or as the holders of the key or code? It was assumed that donors would be asked to give their consent for their medical records to be made available for research, but could this be assumed to be so for their blood and tissue samples? The consensus was that consent for their use had to be explicitly given.

While many who contributed to the workshops were keen to stress that the medical histories that the NHS had kept about

every single citizen over the past half century constituted a unique research resource, the more cautious drew attention to what was happening in Iceland. The genetic structure of the Icelandic people provided a unique source of information about human genetics, given that it was a stable and homogeneous population that had scarcely changed since the ninth century. In addition there were extensive medical records for the population that dated back to the beginning of the last century. A private company, deCODE Genetics, had been given exclusive rights to develop a giant database linking the nation's health records in a non-identifiable form, genealogical background information, and DNA profiles of the entire population of the country.

It was assumed that the citizens of Iceland would consent to the use of their data and tissue. Their government had decided that if citizens did not wish their genetic information to be used by deCODE then they should indicate that they were 'opting out'. Participants to the workshops were uncomfortable with that decision. They took the view that 'opting in' to population research programmes was not just preferable but necessary, and would certainly be essential if we were to enlist the support of the public for large scale epidemiological research.

Mindful what was taking place in Iceland and elsewhere, the UK and the European Union had already taken steps to protect a citizen's genetic data. However, there was a disparity between the collective view of the Union and the views held by some member states, with Europe placing an absolute value on the right of citizens to protect the privacy of their own data. The EU appeared to believe, therefore, that individuals were at liberty to keep their genetic information to themselves or to dispose of it as they saw fit.

INFORMED CONSENT SHOULD BE A FUNDAMENTAL REQUIREMENT IN THE CONTEXT OF RESEARCH STUDIES AS IN ALL FIELDS OF MEDICAL SCIENCE

The principle of 'opting in' should govern the granting of consent in research studies. This should also apply to the use of tissue samples. Participants were not unsympathetic to the public interest argument, but the majority felt that it would be better to encourage people to consent to population research programmes rather than to force them upon them. The idea that people might 'bequeath' their genetic data for research purposes should be explored.

This extreme view did not commend itself to participants. Some felt that there was a public interest in these matters which could not be ignored, that in the last analysis the failure of large numbers of individuals to allow their anonymised or coded medical and genetic data to be analysed would have a negative impact on the conduct of medical research and in turn on the potential for improving our health. The use of identifiable personal information could clearly never be justified without consent, but the public interest might suggest that the use of coded or anonymised information for epidemiological research would be justified if it proved difficult to persuade the public to consent explicitly to its use in research.

Participants were not unsympathetic to the public interest argument, but the majority felt that it would be better to encourage people to consent to population research programmes rather than to force them upon them. The idea that people might 'bequeath' their genetic data for research purposes should be explored. It was also of the greatest importance that the public should be partners in this process. Indeed, if there was ever a subject for detailed public consultation it was surely the whole area of population research and the benefits that would accrue from building up and using large databases containing information on exposures, outcomes and genetic information on individuals.

However, if we believed that there were circumstances in which public concerns should override individual rights, who would decide that the public interest must prevail? It was absolutely essential that the UK should establish a set of principles in respect of confidentiality of medical and genetic data that would balance appropriately the rights of individuals, families and society in the context of both research and clinical practice.

Financial Framework for Health

THE AMOUNT THAT THE UK SHOULD SPEND ON HEALTH SHOULD HAVE REGARD TO THE SPEND ELSEWHERE IN THE DEVELOPED WORLD AND TO QUALITY, AND SHOULD ALLOW ALL INTERVENTIONS DEMONSTRATED TO BE COST EFFECTIVE TO BE AVAILABLE

The 'right' levels of spend should be determined by reference to what was being spent elsewhere in the developed world, to what a reasonable and responsible person might judge to be a service of quality fit for the beginning of the 21st century and

to the resources required to cover all tests and treatments demonstrated to be beneficial and cost effective

Participants in all the workshops considered the resource implications of clinical developments in the wake of genetic science. There was a consensus that the UK should spend an appropriate amount on health and health services, although what might be deemed to be appropriate was less certain. Nevertheless, they argued that that the 'right' levels of spend should be determined by reference to what was being spent elsewhere in the developed world, to an appraisal of what a reasonable and responsible person might judge to be a service of quality fit for the beginning of the 21st century and to the resources required to cover all tests and treatments demonstrated to be beneficial and cost effective.

They also agreed that the services provided should be effective and efficient, responsive and accessible to individuals and result in improvements in health. But if these were some of the key values that should inform future health services, it was equity that they particularly wished to emphasise. There was complete agreement that the services provided should offer geographical, social and personal equity for all our citizens.

Biotechnology had already given rise to a number of cost pressures, particularly for genetically engineered products, such as Factor VIII for haemophilia, human insulin and a growing number of other pharmaceuticals. It was more than likely that with the incorporation of genetic science into mainstream medical specialties other expensive but effective interventions would reach the marketplace and in increasing numbers. No one could be certain what the financial impact of these developments might be, whether they were diagnostic tests or new therapies, but many of the participants were concerned that they might prove too expensive for the UK healthcare system. Over the last two decades other new technologies had ensured that rising costs were a constant factor for health care provision. While there might be individual examples where technology would provide benefits at reduced unit costs, most people were of the view that the cost of health care would inevitably rise.

If permitted, new techniques such as gene therapy, cloning technology or the use of stem cells for a variety of conditions would be expensive. It was less certain what the overall effect would be of the provision of new genetic tests and pharmaceutical agents. The majority view was that despite the anticipated improvements in the efficiency of bringing new drugs to the marketplace pharmaceutical companies would still be obliged to recoup heavy R&D costs in the prices they charged for their products, particularly if we accepted that many new treatments would be individualised for smaller groups of patients. The industry was itself concerned that economies of scale might operate in a negative way if the impact of pharmacogenetics was taken to its limits. In this respect experience had already shown that pharmaceutical companies were reluctant to research and develop products for rare "orphan" diseases.

In the years to come technological developments would be the most important of the pressures on health service costs, of which genetic technologies were likely to provide one of the most challenging examples. Genetic science would lead to novel diagnostic and therapeutic tools, not just those mentioned in the preceding paragraph, but also diagnostic and predictive genetic tests and drugs with novel mechanisms of action tailored to optimise their effect in patients with a specific genotype. There were certain to be other significant clinical applications of this new technology. So how would society deal with these financial challenges while simultaneously meeting the essential aims and values that the workshops had agreed were necessary for the UK?

GENETIC TECHNOLOGIES WILL IN DUE COURSE EXERT ONE OF THE MOST CHALLENGING OF ALL THE COST PRESSURES ON HEALTH SERVICES

In the future the development of genetic science will give rise to considerable cost pressures and provide a unique challenge to society in relation to the funding of health services.

There was considerable support for maintaining the present system of funding for the health service if it were possible to do so. There was much to commend it, in particular for its efficiency and that it commanded widespread public support and affection. There was also a strong and continuing commitment on the part of government to the NHS. But the question that participants kept returning to was whether it would be possible to maintain the status quo in the long term given the challenge of future cost pressures generated by genetic science. Would it perhaps be prudent to consider a future scenario in which the costs of high technology health care might just prove too great for any government to tackle on its own? Might not that combination of social altruism and practicality and a set of values based firmly on notions of equity that had brought the NHS into existence now create new kinds of partnerships between the public and private sectors, with both partners committed to

an agreed set of values in order to reap the health benefits of medical genetics and other new technologies?

While these questions were posed in the workshops, participants acknowledged that the answers to them lay outside the scope of this project and beyond their immediate brief. They were, however, quite firm in their view that whatever the funding mechanisms, resources available to the health care system had to be large enough to allow the full clinical and public health potential of genetic science to be made available to the UK population, subject only to the requirement that the interventions should be cost-effective. A system that failed to deliver tests and treatments of benefit to our population would cease to command the loyalty and respect of an ever expectant and knowledgeable public.

RESOURCES FOR HEALTH SERVICES SHOULD BE ABLE TO MEET THE POTENTIAL OFFERED BY GENETIC SCIENCE

Funding mechanisms must be flexible and the resources for health services able to respond to the requirements of genetic science. A system that fails to deliver tests and treatments of benefit to our population will cease to command the loyalty and respect of an ever expectant and knowledgeable public.

The certainty that we faced difficult choices was as true of genetic technology as it was of other areas in health care, and as much in the diagnostic as the therapeutic arena. Whatever the size of the health budget decisions would still have to be made about priorities. If we were to make the best use of resources then it was essential that we should acquire hard evidence about the costs and the effectiveness of different forms of health interventions. However, the review of the relevant literature that has been undertaken in parallel with the workshops indicated that neither genetic tests nor therapeutic interventions such as pharmacogenetics or gene therapy had been subjected to any form of economic analysis. It was true that health economics had been applied to a handful of areas such as population screening for cystic fibrosis or the thalassaemias, but overall little work has been undertaken in this field.

As participants to a number of the workshops reminded themselves, a health service that intended to make the best use of its resources depended on health economic data. Therefore it was essential to persuade health economists to take a greater interest in issues raised by genetic technologies, and furthermore funding bodies should be encouraged to support research in this field. These issues will be considered in more detail in the next chapter, nevertheless it would seem vital to draw attention to the importance of establishing a properly resourced health economic approach to genetic services.

DECISIONS WILL HAVE TO BE MADE ABOUT PRIORITIES ON THE BASIS OF GOOD HEALTH ECONOMIC DATA

A review of the existing literature has revealed a significant gap in our knowledge of the health economics of genetic interventions. This deficiency must be rectified.

Commercial Considerations

The biotechnology and pharmaceutical industries contribute significantly to the funding of genetic research in anticipation that both health benefits and future profits will be great. Governments and medical charities on both sides of the Atlantic also discern an advantage in funding basic genetic research, so they too make a significant contribution. The funding of the Human Genome Project is one such example. The Single Nucleotide Polymorphism (SNP) consortium, established between the Wellcome Trust, a number of US universities and ten major pharmaceutical companies, provides another illustration of partnership between the charitable and commercial sectors. Partnership arrangements also contribute to the significant funding of basic and applied research in hospitals and medical schools. In many instances academics and employees of the large pharmaceutical companies work together on joint projects, and there are examples of distinguished academics leaving senior positions to take up employment within the industry. Alternatively scientists, with the help of their academic institutions, establish their own start-up companies intending to achieve profits for themselves and for the institutions for which they work.

PARTNERSHIPS BETWEEN THE PUBLIC AND VOLUNTARY SECTORS AND INDUSTRY ALREADY EXIST AND SHOULD CONTINUE TO BE ENCOURAGED

The extremely successful partnerships that have been developed between the public and private sectors in genetic research are an indication of what should be possible in the future.

However, the responsibility and the risks for translating science into commercial products are borne in the main by companies in the commercial sector and by their shareholders. Whatever their size such companies are obliged to return a profit; otherwise they go out of business. This commercial obligation has significant implications for health services. First, decisions about the products that companies seek to develop and the prices that they will charge for them will, at least in part, be determined by commercial considerations. And second, if industry assumes responsibility for and accepts the risks involved in funding, it is industry that will play the greater part in the determining what new products are introduced into the marketplace, and thus how clinical practice may develop in the future. All the participants at our workshops recognised this, and while there might have been some who wished that it were otherwise, none sought to challenge the situation. Indeed, the Policy Exploration Workshop recognised that in order to harvest the fruits of genetic science it would be necessary for government and society to encourage innovation, and to create an environment in which entrepreneurship might flourish and the commercial sector be stimulated and supported.

This support and encouragement would not be without obligations. Industry, in its turn, should accept an agreed set of values that would include partnership, co-operation, commercial integrity and a commitment to equity. Participants in the workshops agreed that it was important to be quite clear about the meaning of 'commercial integrity'. At its simplest it required the pharmaceutical industry to develop cost-effective products that would be of value to as many people as possible. The available evidence suggested that pharmacogenetics might herald a reduction in the number of generic products that had traditionally been designed for a large population base. It would soon be possible to predetermine both the suitability of a particular medicine and the likelihood of developing side effects for individual patients.

PHARMACEUTICAL COMPANIES SHOULD MAINTAIN COMMERCIAL INTEGRITY IN THEIR DEALINGS WITH HEALTH SERVICES.

Pharmaceutical companies must maintain commercial integrity in their dealings with health services. Treatments should remain cost-effective and increased R&D costs should not be used to justify inappropriate profits. Equity should be a key concept. Developing as well as developed nations should have access to tests and medical treatments. The operation of market forces should be balanced against a consideration of the best interests of global public health.

If in the future patients were to avail themselves of individualised rather than generic products, all treatments should nevertheless remain cost-effective. Increased R&D costs should not be used to justify inappropriate profits. Profit levels had to be judged against the values of continuing partnership with health service providers. It was also necessary to include within the notion of 'commercial integrity' the concept of equity, of ensuring that developing as well as developed nations had access to tests and medical treatments. The operation of market forces should be balanced against a consideration of the best interests of global public health.

Partnership between government and the pharmaceutical industry might provide a way of restraining the pursuit of excessive profits. While it was probable that the industry would raise most of the funds required for R&D from its own resources or in the venture capital market, government could assist this process by initiating financial incentives for potential investors while simultaneously giving small innovative research based companies tax exemptions. It was in the long-term national interest that the UK should maintain its current advanced status in the fields of biotechnology and medical genetics. Beyond the ultimate goal of improving the nation's health there were other advantages to be had, for example in employment, in the development of workplace skills, in the manufacture of new instrumentation and, quite simply, in national prestige.

GOVERNMENT SHOULD FOSTER AN ENTREPRENEURIAL SPIRIT WITHIN THE BIOTECHNOLOGY AND PHARMACEUTICAL INDUSTRIES AND ENCOURAGE THE CREATION OF A COMMERCIAL INFRASTRUCTURE TO SUSTAIN THEM WITHIN THE UK

Government has a duty to foster, stimulate and reward the entrepreneurial spirit in the developing field of biotechnology. By providing the infrastructure needed to nurture and sustain its development, it would make it more attractive for industry to continue to flourish in the UK.

In this wider context the pharmaceutical industry relied upon a range of other science and technology based services. The development of genetic tests or medicines needed support in fields as diverse as bio-informatics, robotics, instrumentation and IT. As a global industry it was free to pursue its business in whichever territories it considered were most favourable to its commercial interests, and if it decided to move out of the UK for whatever reason there would be knock-on effects elsewhere in the economy. Government had a duty to foster, stimulate and reward the entrepreneurial spirit in the developing field of biotechnology, and a role to play in persuading the industry to remain in the UK by providing the infrastructure needed to nurture and sustain its development.

THE COST OF DRUGS TO HEALTH SERVICES WITHIN THE UK MIGHT INCREASE.

If costs were determined by what the world market would bear, there could be an increase in the cost of drugs to health services within the UK.

The UK currently managed its national drug costs by negotiating directly with pharmaceutical companies. The possibility that in the future there might be some degree of deregulation in favour of over the counter products could allow these companies to avoid existing pricing systems. Moreover, these mechanisms did not exist to deal with diagnostic products, which were likely to impinge first on health service provision. The internet too would permit the public to deal directly with the pharmaceutical industry in a manner that would bypass most types of local regulation. If costs were determined by what the world market would bear, there could be an increase in the cost of drugs to a health service within the UK, which in turn would be less able to operate what were essentially preferential arrangements in arriving at purchasing agreements that were weighted in the favour of the population as a whole.

How the UK responded to these and to other opportunities, and the framework in which it chose to exercise regulation, would be determined in part by what had been agreed in the EU. In three areas at least, the licensing of new pharmaceutical products through the European Agency for the Evaluation of Medicinal Products (EMEA), the European Directive on Medical Devices, and the European Directive on the Legal Protection of Biotechnological Inventions (98/44/EC), the Union had already promulgated a set of rules governing a number of highly relevant issues. The details of these are discussed elsewhere, but from a purely commercial perspective, what mattered was that the governments of the UK and the other member states of the EU should attempt to move forward together and in step on regulation and related issues.

A number of the workshops considered the commercial aspects of the problem of orphan drugs. They were concerned that the number of persons who might benefit from a certain targeted intervention would be so small that the pharmaceutical industry would decide that it was not worthwhile to develop such products. This might also arise in the case of rare genetic diseases, or as a result of purely pharmacogenetic considerations. It was felt by all the participants that the industry should be encouraged to develop an ethical stance in relation to this issue, and to ensure that those with rare diseases were not disadvantaged.

PROVISION MUST BE MADE FOR SUSTAINING THE PRODUCTION OF ORPHAN DRUGS

The high cost of developing pharmacogenetic products could deter drug companies from investing in research into diagnostics and therapeutics that are only of value to a minority of patients. The industry should be encouraged to develop an ethical stance in relation to this issue and to ensure that those with rare diseases were not disadvantaged. The government in its turn should offer financial advantages for companies to develop such products.

In the United States the Orphan Drugs Act 1982 had attempted to protect patients against such a possibility by offering significant tax incentives to companies who were prepared to develop these so-called orphan drugs. The legislation had defined an orphan disease as a condition affecting fewer than 200,000 persons in the country. The European Commission had since attempted to achieve the same end by implementing regulations for orphan medicinal products. These passed through the European parliament's second reading without amendment and included a market incentive that would allow such products to be freed from the evaluation fees charged by the European Agency for the Evaluation of Medicinal Products (EMEA). This logic could also be applied to subgroups of patients with common disorders segregated by pharmacogenetic considerations. The most effective and safest drugs with fewest side effects might only be suitable for small populations, which would

increase the number of applications for orphan status significantly in the USA and in Europe.

In all of their discussions the workshops recognised that the commercial interests of the pharmaceutical companies were bound up with the question of intellectual property rights, and that developments in genetics had made, and would continue to make this a contentious matter. Even as the human genome was being mapped the arguments had surfaced. To what extent was it possible, or should it be so, to patent a gene or a sequence of DNA? The relevant law in Europe and the UK was covered by the Patents Act 1977, the European Patent Convention 1973 and the European Directive on the Legal Protection of Biotechnological Inventions 1998.

A SOUND FRAMEWORK OF INTELLECTUAL PROPERTY LAW IS NECESSARY TO PROTECT COMMERCIAL INTERESTS

The grant of genuine intellectual property rights was an essential and proper part of commercial activities, designed to protect the interests of those responsible for novel inventions. The pharmaceutical industry should be entitled to benefit from this.

There was little dissent from the view that a sound framework of intellectual property law, which included concepts of copyright, trademarks and patents, was necessary to protect the commercial interests of those who had invested heavily in making discoveries and inventions. It was not the wish of the participants to dissent from such a framework. There was ready appreciation that protection of this sort, together with the corresponding obligation in the case of patents to bring into the public domain the exact details of new discoveries, was an important aspect of the translation of science into technological interventions. The grant of genuine intellectual property rights was an essential and proper part of commercial activities, designed to protect the interests of those responsible for novel inventions. The pharmaceutical industry should be entitled to benefit from any technological advance that translated the pure science of molecular biology into new tests or new treatments.

Nevertheless, many participants felt that the present structure of intellectual property law which appeared to support the patenting of gene sequences was simply morally wrong. The sequence of DNA that made up the gene was a part of an individual's biology, a part of nature, and the knowledge of it should belong to all. It was neither right nor moral that individuals or institutions should seek to gain financially purely from discovering the sequence of bases that made up a particular gene. Others had different reasons for discomfort. For them it was a matter of scientific integrity and part of belonging to a civilised society that a concept as fundamental as a gene sequence should be available to all without payment or hindrance. Some participants were also genuinely perplexed as to how a gene might be capable of being patented given the complexity of existing patent laws and the distinction made there between discovery and invention.

IT SHOULD NOT BE POSSIBLE TO SECURE A PATENT ON GENE SEQUENCES

The present structure of intellectual property law that appeared to support the patenting of gene sequences did not find favour with the majority of participants to the workshops. The sequence of DNA that made up the gene was a part of an individual's biology, a part of nature, and the knowledge of it should belong to all. It was neither right nor moral that individuals or institutions should seek to gain financially purely from discovering the sequence of bases that made up a particular gene.

The position of the pharmaceutical industry was that it was acceptable to seek patents for genes and genetic sequences. The European Directive of 1998 expressly permitted biological material isolated from its natural environment to be the subject of an invention. In the United States genetic inventions were regarded now as indistinguishable from other technological inventions so that the patent claim could relate to the gene, its function or its use. There was a view that the high cost of genetic research and development made it commercially essential to be able to seek a patent on a gene sequence. And that in any case a significant number of patents were granted to academic and government institutions so that patenting did not simply benefit the commercial sector.

THERE SHOULD BE A UNIFORM APPROACH TO PATENT LAW ACROSS THROUGHOUT THE WORLD

Participants also wished to see a uniform approach to intellectual property rights in respect of medical genetics throughout

the world. They were concerned that what could be patented in one country was not always subject of such rights in others.

Participants also wished to see a uniform approach to intellectual property rights in respect of medical genetics throughout the world. They were concerned that what could be patented in one country was not always subject of such rights in others. The European Directive had done much to harmonise the principles by which patents were granted among member states, but there were still differences between Europe and the USA. These differences would need to be resolved.

The Science Base

Mapping the human genome can only be regarded as a beginning. In what has already been called the post-genomic challenge much more will be asked of the scientific effort if it is to be of practical value to society. The genome map that will be rolled out before us will do no more than provide a blueprint for the next stage of the endeavour which is to understand in more detail the underlying nature of biological mechanisms. It is therefore essential to take account of the continuing requirements of the science base in preparing any kind of policy framework for the translation of the science into health benefits.

THE SCIENCE BASE SHOULD CONTINUE TO BE STRENGTHENED

The science base in the UK, already a world leader in the field of molecular biology, should be strengthened. In what has already been called the post-genomic challenge much more will be asked of the scientific effort if it is to be of practical value to society

The workshops chose to identify two aims within this policy area: first, that the science base in the UK, already a world leader in the field of molecular biology, should be strengthened; and second, that at the same time every effort should be made to develop a wider public understanding of the nature and the potential benefits of science and its applications.

All the participants emphasised that science should be regarded not as a cost but as an investment. It was necessary to overturn the longstanding public perception that appeared to embrace the contrary view. As with any prudent investment the eventual benefits would far outweigh the initial costs. In its working practices the science associated with genetics was no different from other kinds of science. A large number of overlapping projects were being undertaken across the country within seemingly discrete disciplines, with funding provided from a range of sources. But in its complexity, there was little to compete with it, and, in its impact, there was little else that had the potential to transform the entire understanding of biological processes and the implications that genetics had for the practice of medicine.

SCIENCE SHOULD BE REGARDED AS AN INVESTMENT NOT AS A COST

We must learn to regard science as an investment and not as a cost. It is in our best national economic interests to sustain and strengthen our existing science base, particularly as it applies to genetic research.

There was general agreement by the scientists in the sessions that the UK government and the research councils should be obliged to take a strategic overview of scientific activities. In the field of genetics, there were complexities of scale, brought about by the need to correlate vast quantities of genomic data with the environmental exposures of many individuals, and the diseases from which they suffered. The statistical analysis required large datasets. The nature of the interactions between gene and environment and their contribution to disease precluded any meaningful results from an analysis of small population groups. The design and establishment of research studies would be both complex and costly. What was required was a managed convergence of research policies and research funding to ensure that duplication was avoided, and that individual research projects were co-ordinated in a creative way.

GOVERNMENT SHOULD TAKE A STRATEGIC VIEW OF THE NEEDS OF GENETIC SCIENCE AND TECHNOLOGY IN THE UK

The UK government and the research councils should take a strategic approach to the research policies and funding needed

for basic genetic sciences and for translational research. In the field of genetics there were complexities of scale, brought about by the need to correlate vast quantities of genomic data with the environmental exposures of many individuals and the diseases from which they suffered.

An increase in the overall level of research funds would obviously be welcome, but there were other imperatives that were thought to be equally important and necessary for the continued success of genetic science and therefore for the improvement of the nation's health. A more strategic approach to translational research and to the development needs of health services was necessary. Basic science provided the elements that would lead to improvements in human health, but for this to happen efficiently and effectively there had to be greater attention to applied research and an appropriate level of resources to support it. In applied, as in basic research studies, there were opportunities for greater co-operation between the universities, the research councils and charities, and the commercial sector.

GENOME VALLEYS SHOULD BE CREATED TO HARNESS THE OPPORTUNITIES FOR PARTNERSHIP BETWEEN THE UNIVERSITIES, RESEARCH COUNCILS, HOSPITALS AND THE COMMERCIAL SECTOR

The success of silicon valleys in the United States and Europe argues for a genetic equivalent within the UK. The government should sustain and support the creation of a number of genome valleys in which genetic science and technology would profit from the interaction between commercial, academic and health service interests.

The example of the Silicon Valley which had given the USA an international edge in the development of IT and associated activities, was frequently cited as a model for the future of genomic science. It was the view of the participants that the strength of the science base would most certainly allow us to create a Genome Valley within the UK. The idea that basic scientists and clinical researchers, academic staff and scientists from industry might all be able to work in close geographical proximity to each other had many advantages. Experience had shown that when these many different elements of the scientific and medical community were enabled to meet and work together we might expect a fruitful symbiosis. Participants argued that there should be a small number of such sites across the nation. Information technology would allow these individual communities separated geographically but serving a single objective to maintain regular dialogue and contact. The aspiration to achieve a co-ordinated set of genetic research activities in the UK would fuel and generate the ideal of a Genome Valley.

As for funding, the present partnership between the public and the private sectors was agreed to be effective and potentially advantageous to both parties. But some participants were concerned that the partnership was still not sufficiently close, that the gulf between the cultures was too vast and that the motives and objectives of the partners were too diverse. It was perhaps important to distinguish between research activity in the basic sciences which might prove useful to the industry and thus attract joint public-private funding, and activities that were primarily concerned with the cost-effective implementation of interventions within the health service where funding should be primarily the responsibility of government. Joint working and joint funding would allow each partner to work towards common gaols while achieving their own objectives, with academics publishing quality research papers, industry providing adequate returns for their shareholders and governments fulfilling their political programmes.

WE NEED A WIDER UNDERSTANDING OF THE NATURE AND THE POTENTIAL BENEFITS OF SCIENCE AND ITS APPLICATIONS.

It will be necessary for society to place greater value on the efforts of the scientific community. The public should take pride in the achievements of British science and understand its contribution to society beyond the purely commercial. The promise of better health that developments in genetics bring with them is a prime example of what science could give to society

There was general concern that the status of those engaged in scientific research had diminished over the past quarter of a century, and that the status of science itself had been eroded. Participants all agreed that it was important to seek to reverse these trends. It was necessary for society to place greater value on the efforts of the scientific community. The public should take pride in the achievements of British science and understand its contribution to society beyond the purely commercial. The promise of better health that developments in genetics bring with them is a prime example of what science could give to

society.

One way of achieving this would be to review the career structure for research scientists and to increase levels of remuneration. This should be accompanied by a public information initiative to encourage pride in scientific achievements, not just in the field of pure science, but as they affected the economic well being of the UK and its status in the developed world. It was also necessary to encourage the young to engage in science and to raise its status as a subject in schools and universities. Good teachers should be encouraged with appropriate incentives, financial and otherwise, to join and stay in the profession.

IMPROVING THE STATUS OF THE SCIENTIST

It is vital to improve the status of the scientist. One way of achieving this would be to review the career structure for research scientists and to increase levels of remuneration. In the longer term it will require a programme of public education about the achievements of British science and scientists, and a determined effort to promote science in schools as an attractive educational option and a worthwhile career opportunity.

There was a view too within the workshops that more needed to be done than simply train geneticists and molecular biologists and to improve their professional status. In the post-genomic age, greater attention would have to given to subjects such as structural biology, protein engineering, pharmacogenomics, biochemistry, and cell signalling, to name but a few examples. Outside the biological arena, bio-informatics, genetic epidemiology, and information technology provided other examples of important disciplines. All would require support to achieve their potential.

VI

Health and Health Service Provision

Introduction

At present genetic services are provided for the UK population through regional genetics centres serving between two and five million people. Many of these centres are linked to university departments and undertake research. They are led by consultant clinical geneticists who with their team of genetic nurses and associates, cytogeneticists and molecular geneticists deliver a full range of genetic services. Traditionally their practice has focused on single gene disorders and chromosomal abnormalities. Much of this work has been linked to obstetric and paediatric practice and to the prevention and management of an individually rare but collectively significant group of genetic disorders. Its focus on the family, and not just on the individual, gives it a unique perspective.

The workload is primarily clinical with much of the effort spent on supporting, advising and counselling patients and their families, and on co-ordinating the management of their disease. There is a close liaison with primary care, with university departments and with colleagues in cytogenetic and molecular genetic laboratories. In recent years, these services have taken on an increasing workload in cancer genetics and medical geneticists have also found themselves spending a significant proportion of their time on developing educational initiatives for the public and other health professionals.

REGIONAL GENETIC SERVICES IN THE UK PROVIDE A MODEL FOR THE CARE OF PATIENTS WITH RARE GENETIC DISORDERS THAT IS ENVIED THROUGHOUT THE WORLD.

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.

This genetic service in the UK provides a model for the care of patients with rare genetic disorders that is envied throughout Europe and the rest of the world. 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 nature of its work has also allowed this service to emphasise its status as a specialised service within the NHS. Despite the difficulties that the specialty faced under the previous administration with its loss of regional designation, the requirement to contract separately with district health authorities and to cross charge for individual tests through mechanisms of extra-contractual referrals, it has emerged relatively unscathed.

However, the service is not without its problems. Resources are constrained while the workload continues to increase. Scientific advances now allow greater potential for genetic analysis yet tests for many conditions for which patients seek help are not available to the service. Planning systems are incoherent and not conducive to the further development of the service. Research grants provide significant funding within the regional centres and pay for some of the tests, but when the grants run out there are no mechanisms by which the tests can move out of the research into the clinical arena.

GENETIC SCIENCE WILL EVENTUALLY LEAD TO CHANGES THAT WILL AFFECT THE ENTIRE PARADIGM OF HEALTH CARE, BUT ALTHOUGH THE PACE OF CHANGE CANNOT BE PREDICTED THERE IS CONSENSUS ABOUT PRIORITIES FOR ACTION

Genetic science will eventually lead to changes that will affect the entire paradigm of health care. Although the pace of change and the timescales for them continue to provoke differing views there is a consensus about the likely sequence of new developments and the priorities for dealing with them. These can be expressed in the form of a tripartite model for the future of genetics within health services.

A Tripartite Model for Future Developments

We have already seen that the new genetics will change fundamentally the way in which we think about health and the manner in which we deliver healthcare: first, by its growing influence within all medical specialties; and second, by permitting a better understanding of its role in the pathogenesis of disease and a more accurate ability to predict and prevent disease. How therefore will these changes impact on clinical services as a whole, and on genetic services in particular? How might these affect the work of the existing genetic services and the practice of medicine in other specialties, both within the hospital and in primary care? Over what period of time will these changes take place? What implications will these changes have for the organisation of health care, for training and manpower planning and for clinical practice? This chapter will focus on the impact genetic science will have on service provision. It will consider the types of service that will be required in the future, the role of genetic testing in health care and its ethical and social implications, genetic screening and a number of specific issues relating to the organisation and delivery of services in the UK.

Participants to all the workshops were in no doubt that genetic science would eventually lead to changes that would affect the entire paradigm of health care, yet, as had been noted elsewhere, the pace of change and the timescales were matters that continued to provoke differing views. These were discussed again at the Policy Exploration Workshop where those present were in the main more cautious. There would of course be individual developments that would affect clinical practice, but large scale major changes were not thought to be imminent. However, a consensus did emerge about the likely sequence of those new developments and what the priorities for dealing with them should be. These were expressed in the form of a tripartite model for the future of genetics within health services.

THE FIRST PRIORITY IS TO DEVELOP A FIVE YEAR STRATEGY FOR THE FUTURE OF THE EXISTING REGIONAL GENETIC SERVICES

The service has tried to evolve in response to scientific developments in genetics and is now under considerable pressure. It is an opportune moment to conduct a review, and to develop a five-year strategy that will, among other objectives, modernise it and reduce existing inequities.

The first priority was to develop and enhance the existing regional genetic service that cared for patients with single gene disorders. The continued support of this service was essential. It could not be ignored, nor its importance diminished in the shadow of the excitement of genetic influences on common complex disorders. The service had tried to evolve in response to scientific developments in genetics and was now under considerable pressure. It was an opportune moment to conduct a review, and to develop a five-year strategy that would, among other objectives, modernise it and reduce existing inequities.

THE SECOND PRIORITY IS TO DEVELOP A HORIZON SCANNING MECHANISM TO MONITOR DEVELOPMENTS PARTICULARLY IN PHARMACOGENETICS AND IN THE USE OF GENETIC TESTS OF SUSCEPTIBILITY

A horizon scanning mechanism that would closely monitor developments in both pharmacogenetics and the use of genetic tests of susceptibility as a means of preventing disease should be developed. It is likely that these developments will have only a gradual impact on health services.

The second priority was to establish as soon as practicable a horizon scanning mechanism that would closely monitor developments in both pharmacogenetics and the use of genetic tests of susceptibility as a means of preventing disease. Participants thought that it was likely that these developments would have only a gradual impact on health services. They also believed that pharmacogenetics would precede and have a greater influence on clinical practice than the use of genetic tests to predict disease risk and allow preventive interventions. This was not to say that within the next ten to fifteen years there would only be innovations based on pharmacogenetic considerations and none which would allow better prediction and prevention of common diseases, but on balance the consensus was that in the early years genetic tests would be more useful in the treatment than in the prevention of disease.

The third priority was to develop specific strategies to deal with the impact of pharmacogenetics and the issues raised by susceptibility genetics. The timing of this activity would be informed by the horizon scanning mechanisms that had been established. These were not unimportant issues, but they represented the longer term future of genetics. The immediate requirement was to establish a process for monitoring future developments that might profit from the fact that it could be some time before it became essential to make detailed plans for these services. We should take advantage of this opportunity to decide what the appropriate strategy should be for them when they are required.

THE THIRD PRIORITY IS TO DEVELOP SPECIFIC STRATEGIES FOR PHARMACOGENETICS AND FOR SUSCEPTIBILITY GENETICS

These represent a longer term future for genetics.

Genetic Testing

GENETIC TESTING WOULD MOST CHANGE OUR IDEAS ABOUT THE IMPACT OF THE SCIENCE ON HEALTH SERVICES

The influence of genetic factors in disease is highly variable and range across a continuum, from one end where they predominate or act as the sole aetiological determinant to the other where they play no more than a minor role in modulating the effect of environmental exposure on disease

The workshops all agreed that genetic testing would most change our ideas about the impact of the science on health and health services. The influence of genetic factors in disease was highly variable and ranged across a continuum, from one end where they predominated or acted as the sole aetiological determinant to the other where they played no more than a minor role in modulating the effect of environmental exposure on disease. For practical purposes, it was possible to distinguish between:

(a)single gene disorders

(b) rare subsets of common disorders due to single genes of high penetrance

(c) common disorders which may be the result of susceptibility genes of low penetrance acting in concert with other genes and environmental factors.

IN COMMON COMPLEX DISEASES IT WILL BE NECESSARY TO DISTINGUISH RARE SUBSETS OF DISEASE DUE TO SINGLE GENES OF HIGH PENETRANCE FROM SUSCEPTIBILITY GENES OF LOW PENETRANCE

These distinctions are not often made but are crucial to an understanding of genetic testing.

Diagnostic and predictive tests for many single gene disorders were already used in the practice of medical genetics. The specific feature of single gene disorders was that the genetic test enabled a diagnosis to be made with certainty. It allowed asymptomatic individuals from families with a history of a genetic disease to be told either that they had the disease and that certain features would eventually appear or that they did not have the disease at all. The discriminatory and predictive power was therefore considerable and had a significant impact on individuals and their families. For example a test would show if individuals from a family with Huntington's disease carried the affected gene. Pre-conceptual testing would also reveal the defective gene responsible for recessive diseases such as cystic fibrosis. Such information allowed couples to make informed reproductive decisions. Testing in the pre-natal period permitted another sort of reproductive choice, a choice relevant to both dominant and recessive disorders. The diagnosis of an affected foetus enabled couples to consider termination of pregnancy. Pre-implantation genetic diagnosis provided a third option for those couples for whom termination of pregnancy was an unacceptable choice. Single gene mutations are also found as rare subsets in a number of complex diseases. Examples include BRCA1 or BRCA2 in breast cancer, MLH1 and MSH2 in colorectal cancer, and PS1 and PS2 in Alzheimer's disease. The

predictive implications were the same as those for the single gene disorders.

Low penetrance susceptibility genes play a significant part in the pathogenesis of common diseases. Their identification would allow us a better understanding of disease mechanisms. But what would be the health benefit of testing for such genes? This was a question that continued to preoccupy participants to the workshops. It was one thing to test for particular genes, but quite another to assume that in establishing their presence clinicians would be able to predict outcomes for their individual patients, or public health physicians for populations. Furthermore would such knowledge lead to the creation of effective interventions that might be able to alter the course of any predicted disease by preventing it from developing or reducing its effects? At their simplest the issues were whether susceptibility testing was feasible and whether it would have a clinical utility.

The optimists were convinced of the benefits that susceptibility testing would bring. In their account of the future, testing promised the ability to predict more accurately the risk of developing common disorders. Test results linked to family history or a knowledge of other risk factors would enable physicians to tell patients which diseases they might be at greatest risk of developing in the future, and the steps that could be taken to lower those risks. These might include surveillance programmes for the early detection of disease, chemo-prevention with drugs, changes in lifestyles to avoid environmental factors that either on their own or in conjunction with the genetic abnormality were known to increase disease risk or in the case of cancer, prophylactic removal of target organs. One participant was prepared to predict that their detection and use in secondary prevention would come about within five years, and in primary prevention within the next decade.

Others were more sceptical. They pointed out that as yet few examples of susceptibility genes had been identified. The genetic abnormalities that were at present linked to complex diseases were in effect rare single gene subsets of those diseases, each with a relatively high penetrance. Although genetic research continued to advance, the correlation between genotype and disease was revealing itself to be a great deal more complex than had been imagined previously. And even if appropriate correlations could be made between certain genotypes and disease there was little evidence that this knowledge in itself would result in interventions that could prevent that disease. It was not at all certain that individuals would wish to have a greater knowledge of their susceptibility to disease, or that, with this knowledge, they would be prepared to take the necessary steps to reduce their risks.

ALL GENETIC TESTS MUST BE FULLY ASSESSED BEFORE THEY ARE INTRODUCED INTO CLINICAL PRACTICE

It is essential for genetic tests to be fully assessed before routine service use. Adequate standards should be set for all aspects of genetic testing, both in relation to analytical and to clinical validity.

The Validity of Genetic Tests

All participants to the workshops agreed that it was essential for genetic tests to be fully assessed before routine service use. The detection of genetic abnormalities was not a straightforward task. In some instances a single gene could be the repository of several hundred different mutations any one of which might result in slightly different clinical outcomes or have no effect at all. In other cases, more than one gene can cause what might appear clinically to be the same disease. Participants were agreed that adequate standards should be set for all aspects of genetic testing, both in relation to analytical validity (that the variants detected are actually present on the gene) and to clinical validity (that the test would be a good predictor of the trait or disease under consideration).

The analytical validity of a genetic test was to a great extent dependent on the skills and experience of the laboratories and their technical staff. Quality assurance was a key factor in ensuring high levels of validity. Participants who discussed these issues felt that within the UK an adequate system of validation and accreditation already existed for tests carried out in cytogenetic and molecular genetic laboratories within regional genetic centres. They were less certain that processes were in place to assure the quality of genetic tests carried out in other laboratories. What the system lacked were mechanisms for the formal evaluation of the analytical validity of new diagnostic tests, and for evaluating new technologies for sequencing genes or identifying particular genetic abnormalities.

Clinical validity was a different issue. It was closely related to the concept of penetrance, which was the probability of developing clinical disease within a given period of time given a genetic defect. Since many classical single gene disorders showed a penetrance of close to one hundred per cent one could say that the existence of the genetic abnormality that it

identified with their presence was a strong indicator of disease. But in most other instances where penetrance fell far below that percentage there was no certainly that the patient who possessed an abnormal gene would actually develop the disease that was associated with that defect. Abnormal traits and diseases were the product of complex cellular mechanisms, which in turn depended upon an interaction between numerous genes and environmental factors. As was observed in a number of the workshops this made it all the more difficult to establish the clinical validity of genetic tests.

Counselling

COUNSELLING SHOULD BE A REQUIRED PART OF THE GENETIC TESTING PROCEDURE FOR PATIENTS WITH HIGH PENETRANCE SINGLE GENE DISORDERS

Every patient who is contemplating taking a genetic test for a single gene disorder should be offered counselling. Such counselling must be supportive and non-directive

There was general agreement in all the workshops that counselling was essential for patients both before and after testing for single high penetrant genes and that it was the predictive power in these circumstances that made it essential. Counselling had to be supportive and non-directive and had to take care to avoid particular courses of action. Before agreeing to a test, patients had to understand their pre-test risks of developing disease and how the test results might alter that risk for themselves and other family members. They would also need to be informed about the range of preventive measures that were available, or, in the case of prenatal testing, of reproductive choices.

By contrast there were mixed views about the role of counselling in susceptibility testing. A susceptibility gene of low prevalence that only conferred an increased risk of disease on an individual and was not deterministic was thought by some to be entirely analogous to other tests used at present in clinical practice, such as the measurement of blood pressure or serum cholesterol. In no instance would the test determine the presence or absence of disease in the future. It would only enable the risk of disease to be defined more precisely. For those participants there was no reason to treat such tests with a greater degree of care than any other type of diagnostic investigation.

Others disagreed. Unlike the benefits that might be gained from the measurement of blood pressure or cholesterol levels, there was at present a huge therapeutic gap between the ability to identify a predictive genetic variant and to intervene successfully. The results of a genetic test would also have a greater implication for other family members. Moreover, pre-symptomatic diagnosis of a genetic condition or detection of an increased susceptibility to a disease might in itself have harmful consequences for the individual, particularly if that knowledge was not accompanied by the ability to ameliorate or treat the condition. The prudent physician would still need to consider with some care the wisdom of carrying out susceptibility testing where little clinical benefit was expected.

COUNSELLING MAY NOT BE ESSENTIAL IN SUSCEPTIBILITY TESTING

Susceptibility testing would only enable the risk of disease to be defined more precisely. It does not have the same predictive value as a test used in the context of high penetrant single gene disorders. There may be no reason to treat such tests with a greater degree of care than any other type of diagnostic investigation.

Genetic Screening

IT IS IMPORTANT TO DISTINGUISH GENETIC SCREENING FROM TESTING

The implications are different and the two are often confused.

Genetic screening had to be distinguished from genetic testing. The differences were of considerable importance and often confused. The media were particularly lax about making the distinction. While appreciating that usage is not totally consistent, our view is that the word 'screening' should be used only to refer to explicit and systematic programmes directed either at whole populations of asymptomatic individuals or at sub-populations in whom risk is known to be increased. Examples would include the neonatal screening programme for phenylketonuria in the UK, and screening programmes for Tay-Sachs disease in an Ashkenazi Jewish communities in a variety of other countries.

SCREENING ASYMPTOMATIC PEOPLE SHOULD ONLY BE USED WHEN WELL DEFINED CRITERIA ARE MET.

We need to understand clearly what we mean by genetic screening of particular population groups and what we intend to achieve with such programmes. In genetic screening, tests may be seen to be imposed on individuals. The ethical dilemmas are magnified and the responsibilities for the physician correspondingly greater. Screening programmes should only be implemented after meeting well defined criteria.

Genetic screening was to be contrasted with "genetic testing". The use of a genetic test on a patient in order to make a diagnosis or to gain a more precise understanding of disease risk was a different matter. Genetic tests were carried out on patients who for whatever reason had taken the initiative and sought professional advice. It was unlike genetic screening where tests could be seen to be imposed on individuals, and in which the ethical dilemmas were magnified and the responsibilities for the physician correspondingly greater. Participants to the workshops all agreed that screening programmes should only be implemented after meeting well defined criteria. In the UK all proposed population screening programmes had to be scrutinised by the National Screening Committee. The American Institute of Medicine had noted that "publicly supported population-based screening programmes [are] justified only for disorders of significant severity, impact, frequency and distribution".

Clinical epidemiological considerations also reveal significant differences between screening and testing. A positive test result from a screening programme would have a lower predictive value than a positive result from symptomatic individuals seeking a diagnosis. This followed from the knowledge that the predictive value of any clinical test was in proportion to the prevalence of the disease in the population in which the test was being carried out. An asymptomatic population had by definition a lower pre-test prevalence or risk of a disease. The application of a test in these circumstances necessarily resulted in a lower predictive value than if the test were applied to symptomatic patients or those who had a greater prior risk. False positive results would also therefore occur more often in a screening than in a clinical context.

Ethical, Legal and Social Issues

Introduction

For many who attended the workshops genetic testing and screening both threw into sharp relief a range of ethical, legal and social issues, and suggested a number of principles that needed to be addressed as genetics became an ever more important part of clinical practice. Participants agreed that pre-eminent amongst these ethical issues for physicians who offered genetic testing to their patients were those of confidentiality and informed consent, both of which derived from a respect for the patient's autonomy. Within the wider social context the issues included the potential impact of genetic information on insurance arrangements, employment practices and the contemporary belief in the right to personal privacy.

Consent and confidentiality had traditionally been the cornerstones on which the relationship between patients and physicians had been built. However, in the view of many of the participants the arrival of medical genetics was likely to affect the way in which we interpreted these tenets and how they operated within best clinical practice. The complexity of many of the issues surrounding, say, genetic testing made it more important than ever that informed consent meant just that. In the genetic future it might be necessary to formulate new guidelines on the nature of informed consent and what it required of both patients and physicians.

THE COMPLEXITY OF MANY OF THE ISSUES SURROUNDING GENETIC TESTING MAKES IT IMPORTANT THAT 'INFORMED CONSENT' SHOULD MEAN JUST THAT

Traditionally, informed consent has been one of the cornerstones on which the relationship between patients and physicians

has been built. The granting of genuine informed consent for testing and for the use of tissue samples in research must continue to underpin that relationship in the genetic future. More than ever physicians will be obliged to ensure that patients understand the nature of the medical intervention that is being proposed.

Human Tissue and Genetic Information

Participants raised particular concerns about the use of blood and tissue samples in research. If tissue was taken for a particular purpose was it ethical to retain it for further tests not specified in the original consent? Was it ethical to store the tissue for future testing for research purposes without the patient's knowledge? The whole matter of human tissue and its custodianship was at present under public scrutiny. How were the rights of individuals to be balanced against the yield of important data from an examination of such tissue that might benefit the health of others? How should informed consent be sought and granted in such a situation? Did the patient have an absolute right to withhold consent for any additional tests and research even if it seemed possible that subsequent research upon them might benefit others within the community? Or supposing that informed consent had not specifically been obtained, or had been obtained only for population based research, and the results of those tests revealed significant information about both the patient and his or her family, should that information be ignored, or did the physician have a responsibility to reveal the test results to the patient, even though it had been taken in the context of a research proposal in which there was no prior intention to relate results to individual identities?

Participants to the workshops then turned to consider these issues in the context of clinical practice. Traditionally a patient's health status had been a confidential matter between the patient and his or her physician. In most cases medical information was only of relevance to the patient. However medical genetics was often concerned with patients in relation to their families or to some wider social group. Mapping the genetic patterns within families by testing and establishing patterns of genetic inheritance could assist the clinician in determining a familial susceptibility to particular disorders. Supposing a patient did not wish to share the results of his or her genetic test with family members, when would it be appropriate for a doctor to breach the traditional bond of confidentiality? In this instance was confidentiality more important than the well being of not one but a number of people? By the same token, did the patient have an absolute right to withhold information from others who might be affected by it? Clearly, in the first instance the physician should endeavour to persuade the patient that sharing this genetic information was in everyone's best interests. However in the event of such a strategy failing it might be necessary in the future to evolve new professional protocols about confidentiality as it touched upon the family aspects of clinical practice.

CAN PATIENTS CONTINUE TO DEMAND ABSOLUTE CONFIDENTIALITY FROM THEIR PHYSICIANS?

The nature of genetic information, the way in which it relates not just to individuals but possibly to members of their families too will call into question the principle of confidentiality that traditionally has governed all dealings between patients and their doctors. In the future confidentiality may be guaranteed to families rather than individuals.

Insurance

Issues relating to insurance were also raised, as they had been earlier when anxieties were first expressed about the danger of genetic discrimination. It required little imagination to see that if an insurance company was to find itself in possession of a genetic profile of a potential client that revealed a susceptibility to a life threatening condition then that company might choose to refuse life insurance or weight the premiums payable in a way that took account of this information. Thus even if the client was able to secure life insurance the cost would be significantly higher that for other individuals whose genetic status remained unknown to the insurer.

ISSUES OF INSURANCE, EMPLOYMENT AND DISCRIMINATION ARE AMONG THE MOST IMPORTANT OF THE SOCIAL AND ETHICAL ISSUES SURROUNDING GENETIC TESTING

Government should have regard to these matters in establishing their regulatory framework.

While considering this scenario the workshops voiced two general concerns. Firstly, given the principal of equity, was it right

for an insurance company to deny cover to any individual or to set premiums beyond their means? And secondly, if that insurance company were to insist that potential clients should disclose their genetic status if they had been tested, or indeed take a test as part of their application for life insurance would this not constitute a form of discrimination? Furthermore an insistence on disclosure by insurers might discourage patients from coming forward for genetic testing in the first place. Thus the issue of insurance was seen by some to be counter to the ambition to improve the health of the general population by drawing down the potential benefits associated with medical genetics.

On the other hand it was also argued that the essence of insurance lay in the calculation of risk - actuarial risk - and that the possibility of developing a life threatening disorder revealed by testing was not so very different from other forms of risk that insurers already calculated in devising policies and setting their premium rates. Why should genetics be regarded as a different kind of risk, particularly when a susceptibility to developing a particular disorder was rather different from an unequivocal prediction of disease?

The participants had noted the concern of the Association of British Insurers in relation to the issue of adverse selection. In their report published in December 1997 *Life Insurance and Genetics: A Policy Statement*, the Association argued that if individuals had access to genetic information which the insurer lacked, then individuals could use this information to secure a better deal than would be the case if information was fully shared. The difference would be made up by the premiums of other policyholders. These issues of adverse selection were discussed by the Human Genetics Advisory Commission in their 1997 report The Implications of Genetic Testing for Insurance. The arguments are summarised in Chapter III.

Employment

By contrast the possible consequences of genetic tests on employment practices in the UK had received rather less attention. In the United States there had been a view that genetic testing might be included in pre-employment examinations in order to screen out applicants who had genuine susceptibilities to harmful reactions from exposure to a particular working environment. It was a matter that had found its way into the medical and occupational health literature. However, a presidential order dated 8 February 2000 now prohibits every federal department and agency from using genetic information in hiring and firing decisions. The President also endorsed the Genetic Non-discrimination in Health Insurance and Employment Bill 1999. This seeks to extend these protections to the private sector and in respect of health insurance as well as employment.

THE USE OF GENETIC TESTING IN THE WORKPLACE SHOULD BE A SUBJECT FOR GREATER PUBLIC DEBATE

Employers should be prevented from introducing pre-employment genetic testing if such testing seemed likely to lead to discrimination and if it were to be used as an excuse for failing to remove environmental health hazards from the workplace

Those workshops that discussed the potential effect of genetic testing on employment practice expressed concern that it might prove easier to test for genetic susceptibility than to remove whatever environmental health hazards there were in the work place. Would this not then constitute an extremely dubious form of discrimination? In any event if employers were to justify pre-employment testing on the grounds that they were seeking to protect a potential employee from unnecessary exposure to risks, tests were unlikely to identify susceptibility to disease with any precision as it might be aggravated by the workplace environment. Indeed it might be the case that employers would use pre-employment tests as an excuse for not ensuring that the environment was safe for all employees. A genuine fear was also expressed that if pre-employment testing was to become general practice then those with high levels of susceptibility might become unemployable. Thus a case of discrimination against an individual might develop into a much broader social issue with significant economic and political implications. It was clear that government should have regard to employment and insurance in their regulatory framework.

Mental Illness

OUR UNDERSTANDING OF MENTAL ILLNESS IS LIKELY TO BE GREATLY INCREASED BY ADVANCES IN GENETICS BUT IT IS UNLIKELY THAT GENETIC TESTS WILL BE OF MUCH USE IN CLINICAL PRACTICE

Genetic research will increase our understanding of the causes for the most common forms of mental illness and in due course lead to more effective treatments. Genetic tests will not prove to be of much use in predicting the onset of common mental disorders given their complex aetiology.

There was broad agreement within the workshops that the effects of advances in molecular biology on our understanding and treatment of mentally ill people merited particular attention. There were clinical, ethical and philosophical issues here that could influence the future provision of health care in this field. The philosophical issues revolved around our definitions of mind and body. There were powerful arguments for resisting what had been dubbed the 'geneticisation' of every aspect of our identities, the reduction of the sum total of human experience including human consciousness to a sequence of genetic interactions. However, this was not to ignore the possibility that genetic factors might play a contributory part in the onset of such conditions as schizophrenia, manic depression, depression, anxiety disorder, obsessive-compulsive disorder and personality disorder.

For the most part the general ethical issues in respect of the mentally ill were the same as those that 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". The particular ethical questions that surrounded the question of reproductive choice were simply thrown into sharper relief in the case of the mentally ill. However, in the matter of ensuring informed consent for testing, treatment or for research purposes there was necessarily a difference when working with these patients. Here the recommendation made in a recent report by the Nuffield Council on Bioethics seemed appropriate. When the patient was lucid his or her consent should be required and sought, and when the patient was in no position to give that consent the guiding principle must be that what was in his or her best interests should be determined by family members and/or physicians.

It seemed to the Nuffield Council's committee that it was unlikely that genetic tests would prove to be of much use in predicting the onset of the most common mental disorders given their complex aetiology. Hence the screening of large sub-groups within the general population for susceptibility to mental disease was particularly unlikely to be helpful. However, a surer understanding of the biochemical processes in human beings as a result of continuing genetic research might lead to the development of more specific and effective treatments for mental illness

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The International Dimension and Human Rights

It is in response to anxieties such as these that a number of international bodies have codified and published their views about the rights of individuals as they relate to developments in genetics indicating what should be regarded as the acceptable limits to genetic research. Examples include the Universal Declaration of the Human Genome and Human Rights that was prepared and agreed upon by the constituent members of UNESCO; the Proposed Guidelines on the Ethical Issues in Medical Genetics and Genetic Services by WHO; and the Convention for the Protection of Human Rights and Dignity of the Human Being with regard to the Application of Biology and Medicine by the Council of Europe. What all these declarations have in common is an absolute commitment to the principles of human dignity, individual autonomy and informed consent and confidentiality in the application of human genetics to medical practice. The WHO guidelines made the clearest statement about what clinicians and healthcare managers should keep uppermost in their minds: "The medical application of genetic knowledge must be carried out with due regard to the general principles of medical ethics, doing good to individuals and families, not doing harm, offering autonomy of choice after information is given, and facilitating personal and social justice". This provides yet another value, that of non-malfeasance or "do no harm" that already constitutes one of the cardinal principles of medical ethics.

Each of these documents seeks to protect the perceived rights of the individual. And in this they do no more than subscribe to the modern view that these are rights that have to be stoutly defended against the encroachment of the state, society as a whole or any overbearing group within society. The same principle is underwritten in the European Convention on Human Rights, soon to be incorporated into British law.

THE PRINCIPLE OF INALIENABLE INDIVIDUAL RIGHTS THAT LIES AT THE HEART OF MOST NATIONAL AND INTERNATIONAL PROTOCOLS AND LEGISLATION ABOUT DATA PROTECTION MAY NOT BE IN THE BEST INTERESTS OF THE HEALTH OF THE POPULATION

Should the individual have an inalienable right to give or to withhold information about their genetic status, particularly if it is encrypted or anonymised, and to prevent their stored data being transmitted to a third party for any purpose including

research when that information might secure health benefits for a large number of other people?

This principle of the all but inalienable rights of the individual lies at the heart of much of the legislation in the UK, Europe, and the United States concerning the protection of data. Individuals are considered to have an absolute right to give or to withhold information about their genetic status, and equally an absolute right to prevent their stored genetic data being transmitted to a third party for whatever purpose. However, one of those purposes might be further research in which individuals' genetic data might assist in securing health benefits for a large number of other people. In such a case should such individuals be able to exercise a right to withhold their genetic information when it might play a part in establishing links and patterns with genetic defects in members of their families or some wider social grouping and thus contribute significantly to their well being? Was there a greater public interest against which these personal rights had to be balanced?

Pharmacogenetics

A number of participants to the workshops voiced the view that pharmacogenetics would be the area in which advances in genetic science would first make its impact on clinical practice. This view was firmly endorsed in the final Policy Exploration Workshop. Pharmacogenetics was the science by which our knowledge of the individual human genome would explain how drugs affected individual patients, and how as a consequence of this knowledge, physicians would be able to tailor interventions to the particular needs of their patients, both in terms of the choice of drug and its dosage. The individualisation of treatment would represent a more refined approach to drug prescribing. At present it was not always possible to predict how a patient would react to a drug, at what dosage level it should be given, how effective it might be, and whether the patient would suffer adverse effects.

It was likely that pharmacogenetics would make a more immediate and potentially greater impact on health services than the use of genetic tests in the prevention of disease. The primary question for service providers would be whether pharmacogenetic products would be affordable and cost effective. Would the prices that the pharmaceutical companies might charge for these new products in order to recoup their investment in R&D be so high as to banish the principle of equity of access to treatment that the workshops had agreed were fundamental to the provision of services? This particular anxiety was countered with the argument that while in the long term the introduction of pharmacogenetic products might well increase the costs of healthcare, the available evidence suggested that individualised drug treatment would lead to a more effective and efficient use of medicines, less wastage, and fewer costs from having to treat the iatrogenic diseases that they caused.

AN IMPORTANT QUESTION FOR HEALTH SERVICE PROVIDERS WILL BE WHETHER PHARMACOGENETIC PRODUCTS WILL BE AFFORDABLE AND COST EFFECTIVE.

It is likely that pharmacogenetics will make a more immediate and potentially greater impact on health services than the use of genetic tests in the prevention of disease. The primary question is whether such individualised products will be affordable and cost effective given the need for industry to recoup their research and development costs.

And would developments in pharmacogenetics dictate a need to reorganise services and change the licensing arrangements for new drugs? A future in which physicians could only prescribe following the receipt of a genetic test result would have logistical implications. The issue of whether the appropriate genetic test would be marketed as part of the treatment or whether the test would be carried out independently of the pharmaceutical company also needed consideration. Would the test requirement be consolidated with the licensing procedures? Might we see a future in which certain drugs would be licensed only for use in patients with the appropriate genotype? These were policy considerations that the workshops were unable to address in detail but which were identified as relevant and important issues.

The Organisation of Health Services

It was clear to the great majority of stakeholders who attended the workshops that advances in the understanding of molecular biology and the translation of that knowledge into health care would have a profound effect on the future organisation and delivery of services. Only a small minority believed that things would continue without change. What concerned the majority

was that there should be continuity between the present and the future shape of services. However, it was clear that such continuity would not occur without planning.

It was unanimously agreed that it was important to build on existing successes. Change should be brought about by evolution not revolution and with a proper regard for the achievements of the past. It was necessary, for example, that the regional genetic services that had served the nation so well since their inception should not be rejected or ignored in favour of a new type of service whose attention was focused on the common complex disorders. The burden of the rare monogenic diseases was large and the bulk of the expertise in the genetic management of these disorders was still to be found within the regional genetic centres. There was a huge educational agenda that was unlikely to be implemented without the full involvement of the existing cohort of medical geneticists, genetic nurses and genetic associates.

THE IMPACT OF GENETIC SCIENCE ON THE ORGANISATION FOR GENETICS LABORATORY SERVICES IS RIPE FOR REVIEW

The Department of Health has already anticipated this by establishing a Working Group on Laboratory Services for Genetics. The report of its deliberations will be of clear value to policy makers in the UK.

There was a consensus too that the impact of the new science on genetic laboratory services was now ready for review. The Department of Health had already anticipated this by establishing a Working Group on Laboratory Services for Genetics. Participants were agreed that many issues had to be considered, such as the number of molecular genetic laboratories that might be required for the future, their siting so as to ensure the widest possible access and efficiency, the nature of future technologies, the relationship between the NHS and commercial sector in genetic testing and the arrangements that would be required in the future for high throughput gene testing. The report of its deliberations would be of clear value to policy makers in the UK.

The participants at the Policy Exploration Workshop had the opportunity also to consider the impact of genetics on a number of recent initiatives within the NHS. It was clear to them that genetic science would be highly relevant to the kind of health service described in *New NHS: Modern and Dependable*. In no sense could one speak of a modern NHS if it failed to take into account the impact of genetics on health services. Partnership was a key theme in that document and equally it was one of the values that was believed by participants to be essential if we were to harness the fruits of genetic science for the benefit of our population. *Saving Lives: Our Healthier Nation* had also emphasised the importance of genetic factors and risk communication in disease prevention.

THE IMPACT OF GENETICS ON HEALTH AND SERVICE PROVISION SHOULD BE TAKEN INTO ACCOUNT IN EVERY NATIONAL SERVICE FRAMEWORK

Within each framework there should be a consideration of genetic influences and of the potential for genetic testing.

The emphasis on quality as a dominant feature of the new NHS was also welcomed by participants. It was essential that clinical matters such as the use of genetic tests, counselling, risk communication and the appropriate use of gene therapy should all be included within clinical governance and have a future place in the National Performance Framework. In view of their wide set of existing priorities, there was some discussion as to whether the National Institute for Clinical Excellence or the Health Technology Assessment programme would have the time and the specialist knowledge to assess genetic interventions. Participants were concerned as to who would take on the responsibility of assessing the validity of new genetic tests or to determine when and by what criteria they would be deemed to be part of routine service provision.

National Service Frameworks (NSF), of which the Calman-Hine Cancer Initiative provided the first example, were also welcomed. It was the view of the participants that within each framework, whatever the subject matter, there should be a consideration of genetic influences and of the potential for genetic testing. Coherence demanded that the organisation of cancer genetic services, for example, should mirror and be consistent with the other services for cancer. This was not to say that in each and every case there would be scope to make recommendations for action, but it was important that the impact of genetic science on each of the subjects of the frameworks should be explicitly discussed and considered. There was no particular demand from participants that genetic services should itself form the subject of a NSF.

THE INCREASED COST PRESSURES BROUGHT ABOUT BY GENETIC SCIENCE WILL MEAN THAT PRIORITIES WILL NEED TO BE SET EVEN WITH PROJECTED INCREASES IN THE LEVEL OF FUNDING

It will be necessary to continue to set priorities and to make choices within a limited budget. This should be publicly acknowledged by politicians, health service managers and clinicians alike. The principle of transparency demands an open and rational discussion with the public. There will be genetic interventions of benefit to individuals that will not be provided by the NHS. The principle of cost-effectiveness requires that medical tests and treatments with a high ratio of benefit to cost are funded in preference to those with a lower ratio.

Resources

How might resources be directed to deal with the impact of genetics on health services? This was a general question to which the stakeholders who attended the workshops continued to return. Current trends in the consumption of healthcare showed that demand continued to outstrip available resources, and there was no reason to suppose that things would be very different in future years. Indeed there was a distinct possibility that in the short term expectations and thus demand would increase more steeply, particularly if there were significant breakthroughs in the field of pharmacogenetics, and if susceptibility testing were to create a new category of 'worried well' patients. These were patients who would not have considered testing, but had been encouraged to act on genetic knowledge that they had acquired from a variety of sources.

It was well understood that resources were finite, and that as far as the NHS was concerned this meant that priorities would have to be set, even with the projected increased levels of funding allocated to the service over succeeding years. There was no dissent from the view that it was necessary to set priorities and to make choices within a limited budget, nor that this should be publicly acknowledged by politicians, health service managers and clinicians alike. The principle of transparency demanded that the issue be discussed openly and rationally with the public and that the fact that there would be certain interventions that would be of benefit to individuals that could not be provided by the NHS should be explicitly confronted. This was what cost effectiveness entailed. Medical tests and treatments that had a high ratio of benefit to cost had to be funded in preference to those that had a lower ratio.

It was to be hoped that demand for services that were less cost effective might be reduced through a process of education that would enable the public to understand that it was in their best interests to moderate their demand in order for services to be made available in response to need. But there was no feeling of certainty among the participants about whether success would attend such a strategy or how these issues would be affected by developments in genetics over the course of the next decade.

Commissioning

THERE ARE PROBLEMS WITH THE COMMISSIONING OF EXISTING MEDICAL GENETIC SERVICES. THESE MUST BE RECTIFIED.

Participants were frustrated that there was not a greater degree of coherence in the systems for planning and commissioning genetic services. The specialised nature of medical genetic services argued for commissioning to be undertaken on at least a regional if not a national basis.

Participants also considered problems with the commissioning of existing medical genetic services. Hitherto they felt that these had been perceived by others to be of less importance and less deserving of funding than disease specific services for cancer, heart disease or mental illness, for example. The commissioning of genetic services had not been a priority for health authorities for reasons that could scarcely be regarded as legitimate. Either they represented too small a proportion of the budget and hence were considered to be less worthy of attention or they were too specialised and therefore not understood. Everyone agreed that these attitudes should be changed and that the growing importance of genetics across the whole of medicine must be acknowledged.

Participants were frustrated that there was not a greater degree of coherence in the systems for planning and commissioning

genetic services. It was difficult to co-ordinate the mechanisms for obtaining capital resources, which were provider based, and for revenue, which depended on negotiations with commissioners. The rules for the cross charging of tests sent between genetic centres were byzantine in their complexity. The structures that previously supported regional leadership and co-ordination had been dissolved and had not been replaced by a cohesive mechanism for planning new developments. There was a great deal of frustration that there was no clarity about exactly which tests or services were to be considered as part of NHS funding and provision, and which were to be regarded as still within the research and development arena. It was essential to make the distinction explicit and to establish procedures by which tests and services at present under development might be assessed to be sufficiently valid and cost-effective to be included within routine services.

WE NEED A MECHANISM TO ASSESS GENETIC TESTS AND SERVICES FOR INCLUSION AS A ROUTINE SERVICE WITHIN THE NHS

There was a great deal of frustration that there was no clarity about exactly which tests or services were to be considered as part of NHS funding and provision, and which were to be regarded as still within the research and development arena. It was essential to make the distinction explicit and to establish procedures by which tests and services at present under development might be assessed to be sufficiently valid and cost-effective to be included within routine services.

It was likewise a point of particular concern that the budget for genetic services was, at least in some regions, to be devolved to primary care groups or trusts. The logistics of agreeing funding between numerous commissioning agencies and the need to seek approval for tests that might only affect a particular primary care group once every few years were too horrendous to contemplate. It was agreed by everybody that the specialised nature of medical genetic services argued for commissioning to be undertaken on at least a regional, if not a national basis. This was one of the strongest messages that should be conveyed to those who had an influence on these processes.

MODERNISATION OF THE NHS WORKFORCE SHOULD TAKE ACCOUNT OF OPPORTUNITIES THAT WILL RESULT FROM GENETIC SCIENCE

New roles and ways of delivering health care that take into account the impact of genetic science should be considered in the review of the NHS workforce.

Human Resources and Manpower Planning

Introduction

It was clear that the integration of medical genetics into current health service provision would make a considerable impact on human resources and manpower planning. Any pressure to postpone the inevitable process of change and to maintain traditional clinical and service practices had to be resisted. The changing roles of health professionals and the modernisation of the NHS workforce were now part of the government's agenda. The onus would fall on all of those who were concerned with health services to explore new ways of delivering care that took advantage of the opportunities offered for improved health by developments within genetics.

GENETICS IS NOT ANOTHER MEDICAL SPECIALITY

Genetics is the core science that underlies all modern biology and embraces the whole spectrum of health and disease. The consequences of the remarkable advances in molecular biology over the past twenty-five years will be experienced in every branch of medicine and medical practice.

Fundamental to the whole process of change and its management was a belief that was expressed in every workshop that medical genetics must not be seen as just another specialty. Genetics was the core science that underlay all modern biology and the whole spectrum of health and disease. The consequences of the remarkable advances in molecular biology over the

past twenty-five years were likely to be experienced in every branch of medicine and medical practice. This was the engine that would drive future health services. Therefore, one should anticipate changes within every medical specialty and consequently the nature of the roles and responsibilities assumed by clinical staff, physicians, nurses and others within those specialties.

Specialist Physicians and Medical Geneticists

WE MUST ENABLE SOME PHYSICIANS IN ALL MEDICAL SPECIALTIES TO GAIN SPECIAL EXPERTISE IN GENETICS

A proportion of physicians in all specialties should gain experience in the genetics. Calman regulations should be reviewed and training arrangements put in place to enable this.

The medical profession had a duty to address the respective roles of the medical geneticist and those trained in other specialties in the light of developments in genetic science. It was clear to all participants that medical geneticists would continue to take a lead in the diagnosis and management of patients with rare monogenic disorders and their families, and that in addition they had a crucial role in the education of other health professionals. What was less clear was how in the future an increasing workload concerned with the genetics of complex diseases would be managed. Was it appropriate that this should be incorporated in its entirety within the workload of medical geneticists? Or was each specialty going to absorb its own genetic work?

THE UK SHOULD PLAN FOR GREATER NUMBERS OF MEDICAL GENETICISTS

It is a matter of the greatest concern that specialist registrar numbers were being reduced. In future years we must plan for an increase in medical geneticists.

Because of its implications for other family members, matters in which medical geneticists were clearly experienced, some participants were of the view that the specialty might only take on the management of those patients within the monogenic subgroups of common disease. Others expressed the view that, since it was unlikely that the medical consequences of genetic science could all be contained within the specialty of medical genetics, it would be necessary for a proportion of physicians in all specialties to gain experience in the subject. Calman regulations should therefore be reviewed and training arrangements put in place to enable a proportion of those training in each specialty to be accredited as a specialist with an interest in genetic medicine. In either case there would be profound implications for the numbers of medical geneticists that were needed in the UK. It would fall to them to carry out much of the training and to provide a body of expertise in the subject. It was a matter of the greatest concern that the number of specialist registrars were being reduced in the specialty. It was inconceivable that we would not require a significantly larger number of medical geneticists, if only because of their role in the training and education of other specialty groups. It was folly to attempt to reduce, or even just to maintain, the numbers in the training grades. The UK should plan for an expansion, not just of medical geneticists, but of clinical professionals with genetics expertise across the entire human resource spectrum.

Genetic Associates and Other Health Professionals

GENETIC ASSOCIATES ARE LIKELY TO PLAY A PIVOTAL ROLE IN THE FUTURE. ARRANGEMENTS SHOULD BE MADE TO ESTABLISH PROPERLY ACCREDITED TRAINING PROGRAMMES AND TO ASSESS THEIR MANPOWER REQUIREMENTS

It will be necessary for genetic nurses and associates to lead the educational programmes and be involved in patient care and counselling. We must increase the number of properly accredited training programmes throughout the country and make a firm commitment to meet their manpower requirements.

Genetic nurses and associates were regarded by a majority of the participants as essential to the service. Genetic associates were likely to play a pivotal role in future years. Participants agreed that neither medical geneticists, nor physicians with special expertise in genetics could respond to the emerging genetic science on their own. It was necessary for there to be a cadre of specialists, among them genetic nurses and associates, to lead the educational programmes and to be involved in patient care and counselling. We had to increase the number of properly accredited training programmes throughout the country, and also make a firm commitment to meet their manpower requirements over the next two decades. The present somewhat ad-hoc arrangements for their training would not be adequate to meet their future needs.

IT WILL BE NECESSARY TO RETHINK THE ROLES OF ALL HEALTH PROFESSIONALS IN THE DELIVERY OF HEALTH CARE IN THE LIGHT OF THE NEW GENETIC AGENDA

We should anticipate changes within every medical specialty and consequently the nature of the roles and responsibilities assumed by clinical staff, physicians, nurses and others within those specialties. Health service managers will also need to understand the implications of genetics.

At the same time it would be essential to devise and to implement in-service training programmes for all health professionals. Such programmes would ideally be multidisciplinary and include basic genetic knowledge, the ethical, social and legal aspects of the subject, and the issues surrounding genetic testing and its impact on individuals, families and society. Planners and service managers should be included within the training programmes, not least because they would need to understand the longer term implications of change and grasp the importance of releasing staff and resources for training as well as planning for inevitable organisational changes.

Public Health Professionals

If the traditional medical paradigm of diagnosis and treatment was about to shift to one that focused on prediction and prevention, then it followed that it would be necessary to include public health professionals within the training programmes. Closer links perhaps would have to be formed between those who at present devised broad based approaches to preventive medicine for the general public and physicians who in future would be suggesting a range of specific measures that an individual patient might adopt to prevent the onset of a disorder for which he or she had been identified as susceptible as a result of genetic testing. University departments of public health would have to embrace genetics and genetic epidemiology and, with the Faculty of Public Health Medicine and other organisations, develop a greater recognition of these subjects within their curricula .

THE UK MUST DEVELOP THE SPECIALTY OF PUBLIC HEALTH GENETICS AND INCREASE THE NUMBER OF PUBLIC HEALTH PROFESSIONALS TRAINED IN THE SUBJECT

Training in public health genetics should be open to a wide range of health professionals, including not only those trained in public health, but also those from a nursing, genetic, epidemiological or other backgrounds. The impact of genetics on health promotion will also be significant.

The development of public health genetics as a sub-specialty of public health was being pursued in a number of places in the United States. The UK should consider doing the same. It was important that such training should be open to a wide range of health professionals, including not only those trained in public health, but also those from a nursing, genetic, epidemiological or other backgrounds. The impact of genetics on health promotion would also be significant and it was essential that those charged with the development of that subject should be aware of the implications for it.

General Practitioners and the Primary Care Team

How primary care might develop in an age of medical genetics seemed uncertain to many of the participants to the workshops. A radical view argued that GPs and the whole primary care system would eventually disappear, that if patients had clear ideas about their genetic status or were generally better informed about medical matters then they might decide to bypass their GP

and proceed straight to some secondary specialist service. The majority of participants felt that news of the death of general practice was greatly exaggerated, that on the contrary primary care would, as it experienced the full force of developments in genetics, find itself in the very front line.

THERE IS GOOD REASON TO SUPPOSE THAT PRIMARY CARE WILL BE THE PATIENT'S FIRST POINT OF CONTACT WITH MEDICAL GENETICS

GPs and primary care teams will need to be genetically literate, fully aware of the ethical, legal and social issues attached to genetics and capable of providing support services such as counselling.

In the first instance patients would continue to consult their GPs. It would be the GP who would administer the simpler genetic tests or read the patient's "genetic chip" without needing to refer patients on to a specialist. This argued that GPs and primary care teams would need to develop a greater understanding of the impact of genetic determinants of health and illness: indeed conversations that related to individual risk would be likely to be located within individual practices. GPs already used family histories in their day to day practice. A more sophisticated understanding of the process of taking such histories, their limitations and the ethical problems associated with them would be needed in the future.

The individualisation of medical therapies brought about by developments in pharmacogenetics would also make its first impact in the GP's surgery. As patients found their own way to information from an ever-growing number of sources, those who worked in primary care would need to meet their patients' knowledge on an equal footing. It was within primary care that the ethical issues relating to confidentiality would appear as of particular concern, although in purely practical terms GPs had long been maintaining appropriate confidentiality in their relationships with their patients and their families.

The impact of genetics on primary care was perhaps one of the most difficult and perplexing areas of discussions within the workshops. For while there was a clear feeling that primary care would play an important role in relation to the future of genetics it was difficult to define how that role would relate to other parts of the health service, and who within the primary care team would assume what kind of responsibility. Would it be the GP? A number of individual GPs were clearly interested and enthusiastic, but the majority were likely not to see the impact of genetics as something of more than marginal importance, given other pressures and priorities. Many believed that it was in this sphere that the genetic associate would find a new and important role and that they would provide the key to the clinical practice of genetics within the primary care setting.

Conclusion

Genetic science seems likely to make unique demands on health care and to change radically the ways in which it is provided. As was stressed in every one of the workshops the future shape of this healthcare and the kinds of demands that it will have to meet are extremely difficult to predict in detail. But that should not become an excuse for ignoring the impact of new technologies on the provision and delivery of services or for failing to plan for the future.

VII

The Political and Policy Machinery

Introduction

POLICY MAKING IN THE UK SHOULD BE PLURAL AND INVOLVE ALL STAKEHOLDERS AND THE PUBLIC

The views of the principal stakeholders and of the general public should be essential elements of policy development. Decisions about future policy can only be regarded as robust when they are informed by the opinions of all who have a legitimate interest in their outcome.

Policy making in the UK has evolved over the past quarter of a century. Whereas it was once essentially hierarchical, with decisions being taken at the centre and then presented to the public for consultation before implementation, the predominant view now is that due weight should be given to the opinions of all those likely to be affected by particular policy decisions before those decisions are taken. The views of the principal stakeholders who have a direct interest in the outcome of what is being proposed, as well as those of the general public who are likely to be affected by whatever is eventually agreed, have become essential elements of policy development. At the heart of the process is a commitment to the principle of full and open consultation, and a belief that decisions about future policy can only be regarded as robust when they are informed by the opinions of all who have a legitimate interest in their outcome.

The participants at our workshops all expressed a commitment to this type of mechanism. Mindful of how the general public had come to distrust genetically modified foods, the workshops were in no doubt that it would be essential to involve them fully in any process that concerned genetics and its consequences for human health; and not just on account of public sensitivities, but because of the complexities of the issues and the rapid advances in basic genetics and molecular biology. The policy process had to command the respect and trust of everyone within the UK. This chapter considers some of the key factors that participants to the workshops agreed would set the context within which discussions about future policy might begin.

Europe

Decisions about the future of health services in the United Kingdom could only be determined in the wider context of policy development throughout the world. As a member of the European Union, the UK would be required to have regard to what was decided in Brussels, its directives and conventions, and to the particular perspectives of individual member states. It was, for example, important that the UK should note the hostility that genetics aroused in Germany, where memories of the evil that had been done in the name of eugenics during the Nazi period inevitably coloured many responses to the subject. While it was quite wrong to suggest that any parallel could be drawn between the abuse of eugenics in the middle years of the last century and the new genetics, we would need to remain sensitive to such concerns.

However, if a policy framework for genetics in the UK was to be informed by what was happening in Europe, it did not follow that European attitudes and policies, as promulgated by the European Commission or in the European Parliament, should be allowed to dictate that framework. Participants to the workshops were adamant that the UK should adopt a proactive stance towards Europe in the matter of medical genetics . The UK was well placed to take a lead on policy for the genetic future of Europe and did not need simply to react to the views of other member states. We had played a dominant role in the Human Genome Project, through the work undertaken at the Sanger Centre in Hinxton, and had developed a world-class research base in genetics and molecular biology over the last few decades. Moreover, the achievement of our regional genetic service in providing for the care of patients with monogenic disorders and their families was a model to which many in Europe aspired.

THE UK SHOULD ADOPT A PROACTIVE STANCE TOWARDS THE EUROPEAN UNION AND PARTNERS IN THE MATTER OF MEDICAL GENETICS

The UK has played a major part in mapping the human genome and its regional genetic services are admired throughout the continent. It is well placed to take the lead in Europe on future policies for genetics.

The Pharmaceutical Industry

Participants to the workshop agreed that pharmaceutical industry would have a major role to play in how genetics would affect health care provision in the developed world. In the field of genetic science the United States had clearly shown a lead, and what was decided in North America would inevitably shape policy in the UK. All the available projections as well as past experience suggested that American health services were likely to be the largest consumer of diagnostic tests and therapeutic interventions developed by the biotechnology and pharmaceutical industries. As a predominantly private service principally funded by insurance, health care in the United States was driven by demand and expectation with some checks on costs provided by the health maintenance organisations. Market forces and the consumer voice were likely to play a much greater role in determining expenditure on health services on the other side of the Atlantic than in the United Kingdom. Therefore it seemed more than likely that the pharmaceutical companies would be able to set prices in North America that would reflect their heavy investment in research and development, and at a level well beyond what would be regarded as affordable by our own health services.

The very different ways in which the United States, the UK and Europe fund and organise health services have provided a rich and diverse field of operations for the pharmaceutical industry. Traditionally self-interest had determined its decision to base its operations within the UK. The unique role of the NHS as the principal provider of health services with a relatively straightforward and common sense attitude to the protocols that should govern the clinical trials of new drugs enabled the industry to test and launch products faster and more efficiently in the UK than elsewhere. As part of Europe the UK was also an important commercial bridgehead into one of the world's largest markets for health care products. The price that the industry paid for these perceived advantages was the special arrangements that they were prepared to negotiate with the NHS about the supply of pharmaceutical products to the service. In return it had been understood by all parties that any licensed pharmaceutical product could be prescribed by any medical practitioner without government interference.

Recent years have seen an erosion of these freedoms. Cost pressures within the NHS have led individual hospital trusts and practices to restrict the range of products they use. The establishment of the National Institute for Clinical Excellence was thought to be working to the same agenda but on a national level. It was entirely possible that in the future disagreements would emerge between the NHS and the industry on the range and the price of drugs that the NHS would purchase, and if it was unable to sustain an appropriate level of profitability in the UK in relation to the advantages that it gained from partnership with the NHS, the industry might seek to withdraw from the pricing arrangements with the NHS and remove its manufacturing capability or its research base outside the UK. If the future just outlined above were to come about, the consequences for UK science and for its economy would be grave indeed. It was therefore essential to devise future arrangements in such a way that they would be mutually beneficial for all parties.

IT WILL BE ESSENTIAL TO MAINTAIN THE EXISTING PARTNERSHIP BETWEEN THE PHARMACEUTICAL INDUSTRY, UNIVERSITIES AND THE HEALTH SERVICES

Science in the UK and the national economy is heavily dependent on partnership between government, academia and the commercial sector. It will be essential to devise future arrangements in such a way that they will be mutually beneficial for all parties.

The part played by biotechnology and pharmaceutical companies in funding research in genetics in the UK was agreed by participants to the workshops to be vital in determining future policy in the UK. Traditionally the funding for science had come from both the private and the public sectors. The interests of both were served by these arrangements. The nation benefited economically by way of the investment potential and employment opportunities and directly from the impact of the science itself. Industry in turn benefited from partnership arrangements with scientists and physicians in the best of our universities and hospitals. A future scenario in which these arrangements ceased to be of value to either party and that allowed the industry to reduce or withdraw its investment would have a significant impact on the ability of the UK to play a leading

role in genetics and molecular biology. Several of the workshops gave considerable thought to this question, and while arguing that that co-operation between the public and the private sectors was in everyone's best interests, they agreed that government would need to take a lead in setting policy guidelines for genetic research and to perpetuate an environment in which partnership was understood to be the key value in funding all kinds of research.

THE IMPACT OF GENETIC SCIENCES ON RESOURCES WILL BE ONE OF THE MOST SERIOUS CHALLENGES YET FACED BY THE NHS

The enormous task of reskilling the vast workforce of the NHS, the cost pressures of new technologies and managerial inertia may prevent the NHS from adapting itself to a new kind of medical practice. There is huge support for maintaining the present system of funding, but it will be necessary to ensure that resources will be available to enable all citizens of the UK to benefit in the future from all genetic tests and services which are shown to be cost-effective, and that the system will be sufficiently flexible to adapt easily to changing technologies and patterns of service delivery.

Funding Issues

Participants to the workshops were encouraged to 'think the unthinkable', and they agreed that perhaps the gravest challenge ahead might be that the NHS as a national health delivery service would not survive the introduction of new technologies brought about by a greater understanding of biological mechanisms. The enormous task of training and educating so large and so varied a workforce, the cost-pressures of new technologies and managerial inertia in the face of overwhelming change might quite simply defeat the present system. But the majority of those who attended the workshops were emphatic in their support for the present service and its achievements, the values that it espoused and its principles, notably the ideal of health care for all free at the point of delivery. They wished to maintain the present system of funding, but also to ensure, first, that resources would be available to enable all citizens of the UK to benefit in the future from all genetic tests and services that were shown to be cost-effective; and second, that the system would be sufficiently flexible to adapt easily to changing technologies and patterns of service delivery.

It was not within their brief to consider how health services within the UK were to be funded in the future. However they were all quite clear that services should be provided within the framework of agreed values, and that it was the responsibility of government and of the regulatory framework to ensure that those values were maintained.

The Role of Government

Government has a direct role to play in the determination of health policy and in its implementation. The Department of Health, and the NHS Executive within it, has responsibilities ranging from liaison with outside bodies and organisations on all manner of health issues to the development of policy for health services and its implementation. The Secretary of State for Health is accountable to Parliament for his stewardship of the nation's health and its health services. The health service in the UK is provided and funded by the state.

But what did we mean by 'government' in this context? It was clear that within government there were a number of different stakeholders each with a interest in the developing field of genetics: the Scottish Parliament, the Welsh Assembly, the Department of Health; the Scottish, Welsh, and Northern Ireland Offices; the Department for Education and Employment; the Department of Trade; the Office of Science and Technology; the Department of Transport and the Environment; and last, but by no means least, the Treasury. In addition the Policy Unit within the Prime Minister's Office and the Cabinet Office, through its Performance and Innovations Unit, had an interest in developments in human genetics.

A view expressed in a number of the workshops was that each of these interested parties looked at the genetic future from their own perspective, a perspective that to a great extent was determined by their own political and administrative brief. Thus the Department of Health was chiefly concerned with the policy impact of genetics across the whole field of health, including such matters as new reproductive technologies and biotechnology and its ethical and legal implications, and more specifically their impact on the provision and delivery of health services in England. The Scottish Parliament and the Welsh Assembly held the briefs for the devolved delivery of health services within their respective countries, while the Northern Ireland Office was directly responsible for health services in the Province. The Department for Education and Employment was interested in the likely effects of developments in genetics and biology on general and vocational education at every level from primary to tertiary, and with future training requirements. The Department of Trade and Industry was there to encourage commercial

development, and thereby to secure the economic future of the UK. Its priority was to engage with global players to establish or maintain an existing British presence, while also to foster smaller local operations in order that the UK would remain at the very forefront of the bio-technologically based industry. The Office of Science and Technology as part of the DTI championed the cause of science and investment in the nation's science base and the work of the various research councils. The Department of Transport and the Environment had the responsibility for environmental policies, and for land use planning within the regions. The Treasury's role was to mediate during the annual public spending round, making political judgements about what programmes should be funded and at what level.

Somewhere within this nexus of overlapping interests a number of extremely important decisions would have to be taken as we planned for the future. Who would decide which services and which patients were to be funded? Who would fashion the ethical framework that would be required for the practice of medical genetics and where would ethical issues be resolved? Who would set the research agenda for future work in molecular biology and for the development of new medical interventions? If government failed to give a lead then it would be left to market forces to provide the answers to some of these questions. The workshops were in agreement that laissez faire was no substitute for properly conceived planning.

A large proportion of the participants to the workshops were also in agreement that what was lacking in government was any kind of co-ordination of the different ambitions and perspectives of each the interested departments of state. There was an absence of what had been called 'joined up' government. It was observed on a number of occasions that following on from the devolution of political power to Scotland and Wales, and perhaps Northern Ireland too in the future, the responsibility for health services outside England had been relocated from the Secretary of State in Whitehall to Cardiff and Edinburgh, and that the Prime Minster was now the only member of the UK government who spoke for the whole nation on all issues, including health. Was the co-ordination of policy perhaps a task that should naturally fall to the Prime Minister's Office, or to its Policy Unit? Or should a responsibility for policy co-ordination throughout the UK be delegated to, say, the Department of Health?

THERE MUST BE JOINED UP GOVERNMENT

All government departments should co-ordinate their particular departmental interests in genetics. The role of co-ordinator might fall naturally to the Prime Minister's Office or to its Policy Unit. Alternatively the Department of Health might take on that role on behalf of the UK government. For the Prime Minister's Office to lend its authority to this development would also enhance the UK's status within the international community and increase the kind of political leverage we could expect to deploy in protecting the interests of the UK and its citizens.

But who would actually drive the making and the implementation of genetic policy? Co-ordinating a range of government stakeholders was one thing, taking account of international commercial ambitions in the field of biotechnology, matching European aspirations to British intentions, and harnessing the enormous power of the science base in the United States to the interests of the UK was another. These activities required a very particular kind of political authority that drew its strength from the knowledge that it spoke and negotiated for the whole country. Many of the drivers of genetic change lay beyond the control of the UK, but to employ a common metaphor, we might be able to ride them for our best purposes rather than let them chase us hither and thither. To do that successfully, concluded the workshops, would depend on establishing a policy framework that was stamped with authority and driven by a single minded political will to succeed.

THE LACK OF AGREEMENT ABOUT THE PACE OF DEVELOPMENT AND TIMESCALES WILL MAKE IT MORE DIFFICULT FOR POLICY MAKERS

The failure of experts to agree on this issue of pace places policy makers at a disadvantage. They will need to know what might be achieved within an agreed time frame.

Scientific Drivers

One of the chief difficulties that attended the devising of such a policy framework was disagreement about the timescale within which research in molecular biology would be of use in clinical practice and thus bring benefits to human health. Within the workshops two views were expressed about the pace of scientific development. While everyone acknowledged the remarkable achievement of mapping the complete human genome and concluding the project well in advance of earlier

expectations, different views were expressed about the pace of future development. Some were doubtful that in the post-genomic period progress would be as swift or as smooth. The complexities of the science would result in progress being in fits and starts over many decades. Others took a more optimistic view. They argued that genetic science would continue at its current pace and that aided by new technology, by ever more flexible information systems and refinements in bio-informatics, the ability to transform this growing knowledge into practical healthcare would keep pace with the scientific research. These participants were certain that significant benefits would be seen over the next decade. However the failure of experts to agree on this issue of pace placed policy makers at something of a disadvantage since they needed to know what might be achieved within an agreed time frame, even though no one could predict with any certainty what might happen and when.

Values and Principles

The uncertainty about exactly how and when the science of genetics might impinge on clinical practice or on the potential for preventing disease seemed to many who attended the workshops to reinforce the case for policy development being grounded in and informed by an explicit and agreed set of values. A number of values emerged from these discussions. First, it was agreed that services should be equitable, effective, affordable and of high quality; second, that the relationship between all parties should be guided by principles of partnership and co-operation; and third, that the process of agreeing policy should be informed by the values of transparency and integrity.

AT THE HEART OF A POLICY FRAMEWORK FOR MANAGING THE IMPACT OF MEDICAL GENETICS ON UK HEALTH SERVICES THERE MUST A SET OF AGREED VALUES

The values that should underpin a policy framework for planning the introduction of medical genetics into UK health services should include equity, effectiveness, affordability and quality. Relations between all stakeholders should be grounded in partnership and co-operation, and the process of agreeing policy should be informed by the values of transparency, or openness, and integrity.

Values and Principles

- Equity
- Effectiveness
- Affordability
- Quality
- Partnership
- Transparency
- Integrity

Participants were concerned to elaborate on what they meant by equity in the context of genetic services. Broadly speaking it was deemed to include equity of geographical access, that whatever genetic services were available they should be equally accessible across the whole of the UK, and equity in terms of disease status, in that the patient suffering from a rare condition should not be disadvantaged purely because it was rare. In these terms it would be thought inequitable that services for common diseases were funded to higher standards and were more easily available than services for a rare disorder.

Quality was a value that related to every aspect of health services from clinical practice to their organisational systems. A proper understanding of what quality meant was the best way of ensuring that adequate standards were attained throughout health care, and as far as genetics was concerned it would play an important part in evaluating novel tests and therapies.

The workshops identified effectiveness as an important value in respect of the delivery of health services and the kinds of intervention that we might anticipate from advances in genetics. Effectiveness meant that services and interventions should achieve the objectives that were agreed for them. As for affordability, that was essentially an issue of cost-effectiveness. It was not particularly helpful to develop a treatment that was so expensive in relation to its effect that it could only be made available to a few and thus its benefit in relation to its costs were small when compared to other interventions.

Transparency, or openness, should be regarded as essential in all relationships in healthcare: between patients and doctors,

between clinicians, associated medical staff and service managers, and between the health service itself and the general public. Equally all dealings between politicians and healthcare staff and the wider public should be entirely transparent. It must also be understood that all of these relationships were partnerships. The ideal of partnership between all interested stakeholders should determine both the nature of the relationships and the way in which they were conducted.

THE PUBLIC ARE PARTNERS IN THE POLICY PLANNING PROCESS

The lesson to be learnt from the reaction to genetically modified foods is that the public has a right to be a partner in all decisions about policies in this area. What is required is an educational dialogue between all parties.

The Public

It was a common theme and a matter of total agreement that members of the public should play an equal part in the kinds of partnership that were necessary to develop policy in a legitimate manner. However the public would need to know a great deal more about genetics if they were to participate fully in the discussions. If the workshops kept returning to the debate that had been raised about genetically modified foods that was because they saw in it a potent warning about how poorly versed most people were about the whole subject of genetics and how lack of understanding and knowledge bred suspicion and prejudice. It was also because they realised that, irrespective of their technical knowledge, the public had a right to express their concerns and fears, and that policy makers had a duty to take note of theses. It was important too that the media were properly briefed in order to mediate complex scientific subject matter as well as developments in medical technology fairly and accurately for their viewers, readers and listeners. The need for a programme of public education had already been identified as one of the key policy areas. At issue here was how such a strategy should be devised and implemented and who was to lead and be responsible for it.

THE PUBLIC HAVE SPECIFIC AND LEGITIMATE CONCERNS ABOUT GENETICS THAT MUST BE TAKEN SERIOUSLY

It is essential to recognise the kinds of views that exist around the subject of genetics and to acknowledge the publics genuine fears and concerns.

Involving a public who would be better informed about genetics in the planning of this policy framework was never an issue for any of our participants. What did concern a number of them were the mechanisms that might be available to ensure that any process of public consultation was both efficient and effective. A range of options that included citizens' juries and focus groups were considered. The use of opinion polls to gauge public attitudes was also thought to be of value, particularly in charting shifting public opinion. Here the need to increase public awareness and understanding of human genetics and the need to measure attitudes to aspects of the subject would appear to come together. A flexible programme of public education would be required to meet the changing information requirements of the general public as revealed by polling, to remedy misunderstanding and to dispel inaccurate and misleading information.

The Profession

It seemed likely to many of those who participated in the workshops that some aspects of clinical practice were likely to change more than others as a result of the introduction of medical genetics into health services. Primary care was often cited as one of the settings on which genetic science would have a significant impact. Primary care could either shrink or disappear as patients decided to go straight to specialists, or become the first and most important point of contact between doctors and their patients. Whichever scenario you subscribed to every workshop acknowledged that patient-doctor relationships were about to change. In the future patients were likely to treat with their doctor on a more equal footing that had often been the case hitherto.

THERE SHOULD BE NEW MECHANISMS FOR LIASING WITH THE MEDICAL PROFESSION AND RESEARCH SCIENTISTS

The medical profession in its widest sense, doctors, nurses and allied health workers as well as research scientists working in the field of genetics must be included in the policy planning process. It may be that it will require new mechanisms such as a new 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.

This evolving relationship made it important that the mechanisms that enabled the formulation of public policy should take account of the interests of the physician and other clinical professionals. Clearly the profession in its widest sense, doctors, nurses and allied health workers, would need to be included in the planning process. There might well be a need to develop new mechanisms for liasing with the professions at national, regional and local levels in order to ensure wholehearted co-operation in planning for the future. The same would be true for research scientists who in a local sense were one of the drivers of change in the provision of health services, whether as pure scientists working in molecular biology or as technologists developing clinical products from that research. Possibly we should be anticipating a new set of relationships between pure and applied science, clinical medicine and the planning process, all of which would demand flexibility. What was perhaps required was a joint policy forum on academic and innovative medicine that would consider and debate the impact of scientific developments in the field of genetics and its impact on medical and public health practice.

Recommendations and the Policy Framework

Introduction

The majority of the workshops assumed that the NHS was central to the whole of the planning process and that it would continue to be the principal health provider in the UK. Some participants were tempted to conclude that existing planning mechanisms for health policy in the UK should be swept away; that since medical genetics represented such a break with past practice at every level it offered a rare opportunity to wipe the slate clean and to start afresh. However, the view that commanded the most support through all of the workshops was that there was already a range of structures in place that could be adapted to and adopted for the creation of new policy.

In terms of health service delivery the Department of Health and the NHS Executive had a range of existing advisory committees, such as the Human Genetics Commission, as well as departmental groups who might play a valuable part in helping to plan for the future, as did the Department for Education and Employment and the Department of Trade and Industry. Then there were the Select Committees in the Houses of Lords and Commons, and the Joint Genetics Committee of the Royal College of Physicians, the Royal College of Pathologists and the British Society of Human Genetics. It was a matter of co-ordinating these existing committees, groups, bodies etc as they looked to the future. But how should they look to the future?

The Policy Framework

The workshops recognised that prediction, whether of ultimate outcome or pace was not a particularly worthwhile activity. Attempting to second-guess what might come about and when was thought by most to be futile, and more likely to be wrong than right. However it was essential that policy makers should understand the issues before them and prepare for, influence and respond to them. But such understanding and preparedness could not be cultivated in a policy vacuum. Equally, there had to be some points of reference against which the scientific advances and the views of the public could be assessed. And these points of reference might constitute a framework within which policy for the future could be formulated. However this policy framework should not succumb to the temptation of trying to predict the future.

The policy framework would have to be sufficiently strategic and flexible for its policies to be adapted if confronted by unforeseen threats and reverses, as well as taking full advantage of unexpected opportunities. It would consider the totality of health service provision in the UK and not simply that which might be provided by the NHS. All of the key policy areas indicated earlier should be addressed, together with the implications for health services and the nature of the policy process itself. At the same time it would be important to take the widest possible view of the benefits that might accrue to the UK from advances in genetics, recognising the contributions made by science and medical technology to the national economy in terms of employment, revenues and prestige.

It was the view of the participants that policymaking within this framework had to be rooted in a set of core values. They did not wish to be prescriptive about the nature of these values, but there was general agreement that principles such as equity, responsiveness to the individual patient, accessibility, quality and cost effectiveness were all-important and should underpin decisions that related to the development of genetics within the UK. Values were particularly important in situations of uncertainty. Their existence could provide a way of resolving specific problems in a fair and transparent manner. In relation to equity there was a particular concern that the impact of genetics on healthcare should be considered in a global context, that the existing inequities between the developed and the developing world should not be widened as a consequence of the new technologies but that they should be narrowed; and that in relation to the development of new therapies there should be regard to the morbidity and mortality caused by those diseases prevalent in the developing world.

Partnership would be paramount. The nature of scientific endeavour required many different groups to work together. The commercial sector, governments, the research councils, scientists, health professionals, and the general public all had a stake in the subject. However it was important to remember that each partner necessarily had different objectives. Tensions were

inevitable, but taken in the context of a national policy framework that had been agreed by all the stakeholders, it would be easier to make those compromises that would in due course benefit everyone.

Principal Recommendation

The UK government should take the lead in developing a policy framework that would provide a context within which scientific developments in genetics and molecular biology and their clinical and public health applications might be assessed. The framework should:

(a) be informed by a set of core values

(b) take into account the issues set out in this report

(c) be developed in consultation with all relevant stakeholders and with the public

(d) ensure partnership arrangements between industry, the universities and the health service

(e) emphasise the need for co-ordination across all government departments

Policy Recommendations

The Regulatory Framework

1.The UK should agree upon a set of principles which would underpin a regulatory framework that must be light enough to encourage promising developments in genetics but rigorous enough to protect the public. The regulatory framework should:

(a)be based on values and principles which apply across both public and private sectors and throughout the UK

(b)enable the UK to take full advantage of research and development

(c) protect the health interests of the public

(d) have regard to international trends and developments

(e) be able to arrive at decisions in a timely and expeditious manner

(f) be able to impose moratoria on activities that were thought to be ethically or medically unacceptable

Educational Strategies

1. The UK should establish and implement a strategy for the promotion of genetic literacy. The strategy should:

(a)consider how a concerted campaign might be mounted to raise public understanding of genetics and the issues surrounding personal risk

(b) ensure that genetics and education about risk would be included in the national curriculum and in the curricula of all relevant disciplines in our universities

(c)include a systematic approach to the training and education of health professionals

Information and Confidentiality

1. The UK should establish a set of principles in respect of the confidentiality of medical and genetic data that should cover both clinical and research activity. The principles would aim to:

(a) balance the rights of individuals, families and society

(b) ensure that all personal health information continue to be protected by the full force of law

(c) consider the public interest in the use of coded or anonymised data in research studies

(d) emphasise the use of informed consent as a fundamental requirement in research studies

Financial Framework for Health

1.The UK should have regard to the cost pressures posed by the impact of genetic technologies on the health service and ensure by whatever means the necessary resources for the funding of cost-effective interventions arising out of genetic science.

2.The UK should encourage the use of health economics in assessing the impact of genetic science on health services

Commercial Considerations

1. The UK should support the enterprise culture, foster an entrepreneurial spirit within the biotechnological and pharmaceutical industries, encourage the creation of a commercial infrastructure responsive to their needs and continue to use fiscal and other incentives to stimulate partnerships between them and academia. In return it should require of the industry a commitment to commercial integrity in relation to:

(a) the costs of drugs to the health service

(b) equity of access to drugs both within and without the UK

2. The UK should consider further the issue of the patentability of genes and genetic sequences, and whether and how it might be possible to preclude the patenting of genetic sequences while permitting individuals and institutions to reap the financial benefits of inventions and products.

The Science Base

1.The UK should consider further investment in its science infrastructure and actively encourage the development of a cluster of genome valleys.

2.The UK should devise policies to arrest the decline in the status of science in society by reviewing and augmenting the career and salary structures of scientists and teachers of science, and by explicitly encouraging and improving the teaching of science in secondary schools and universities

Health and Health Service Provision

1. The NHS should develop a five year strategy for regional genetic services which would among other objectives aim to reduce existing inequities in the service. The strategy should:

(a) take into account the recommendations of the Working Party on Laboratory Services for Genetics

(b) establish a mechanism for assessing the validity of genetic tests before their introduction into clinical practice

(c) take into consideration the social and ethical aspects of genetic testing especially in relation to employment, insurance and discrimination

(d) establish new and coherent mechanisms for the development and commissioning of medical genetic services

(e) review the responsibilties of medical geneticists in the light of their increasing workload and role in education and training and consider the implications of these responsibilities in terms of manpower requirements

(f) review the recruitment of genetic associates and the arrangements for their training and accreditation

2.The UK should establish a 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.

3. The UK should aim to establish at some future date detailed strategies on the impact of pharmacogenetics and of susceptibility testing and prevention on health services but should consider at an early date:

(a)the roles of health professionals and their needs in the light of the new genetics as it impacts on health services

(b)how a proportion of physicians in every specialty might be trained and gain experience in genetics and be formally accredited as having special expertise in the subject

(c)the development of public health genetics as a discipline for public health professionals and its educational implications

(d)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.

Postscript

If anything can be said with certainty about current developments in genetics it is that they are highly unpredictable. Therefore much of the material in this report has been necessarily conjectural. However, it is conjecture informed by the best available opinion and sustained by expertise. The report has attempted to gather together and give a shape to the opinions and arguments of men and women working in healthcare in the UK who have already thought deeply about the whole subject area of genetics. Therefore it should be read as a report of what seemed, and indeed continues to seem important as we approach a time when all medicine in the UK will take its clinical cue from the extraordinary advances in human biology that occurred in the closing years of the twentieth century, and as we begin to anticipate a quite new series of demands on health services in this country.

Taken on their own there is little that is entirely novel about the individual issues that were raised in the sequence of nine workshops held in the summer and autumn of 1999. As the brief bibliography attached to this report makes plain all of these issues from the human biology that underpins medical genetics to the raft of ethical, legal and social questions provoked by likely changes in clinical practice as a consequence of this biology have already been considered in depth by a range of professional commentators on both side of the Atlantic. But then this report was never intended to be read as a *vade mecum* of what is happening in the field of human genetics or what the lie of the land might be beyond that border that separates the present from the future.

Its virtue, we believe, resides in the fact that the workshop process allowed us to draw together a remarkable range of well-informed contributions, to compare them with each other and against a range of models for future health care services, and then to organise them into seven over-lapping policy areas. As far as we can judge this is the very first time that anyone has attempted to take a view of how the many issues provoked by the biology that underpins medical genetics relate to policy decisions to deliver health benefits to the population of the UK. It is this that gives the recommendations in this report their particular force.

What unites those individual recommendations is a single overarching recommendation, namely that the United Kingdom should develop a broad policy framework which can underpin all discussions about future policy in respect of medical genetics and its impact on the future provision of health services. It is worth repeating that it was the view of all of the workshops that this was a task that could not be postponed. If we take the popular and the dramatic view of the likely effects of genetics on health services in the UK, the tsunami, the tidal wave, is only just below the horizon and it is rolling towards us at an ever-greater speed.

Reports such as this are sometimes written to gather dust. They are published; public and professional opinion burns brightly for a while; possibly their recommendations are referred to an existing or to an ad-hoc committee for scrutiny; and then they fade into oblivion. It would be a disservice to all of those men and women who contributed their time and their expertise so generously to this project if that were to be the fate of this report. More important it would fly in the face of the hopes that were expressed most notably at the final Policy Exploration Workshop.

Participants to that two-day event were quite clear that words should be followed by deeds and not just more talk. If the report of the whole project could be regarded as a broad statement of principles, these principles must be accompanied by an action plan that would set out unequivocally what needed to be done and when it should be done. Naturally the responsibilities for both devising and implementing such a plan lie beyond the remit of the Nuffield Trust or the Public Health Genetics Unit. It is government in partnership with a full range of stakeholders in the new genetics that should take up this responsibility.

For their part the Nuffield Trust will host a meeting in the Autumn of 2000 for all those who took part in the Policy Exploration Workshop together with other relevant contributors in order to establish an action plan based on the recommendations of this report. In the meantime the Trust intends to monitor what action is taken in respect of these recommendations in the short and the longer term.

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