



Government Response to the
House of Lords Science and Technology
Committee Inquiry into
Genomic Medicine

Presented to Parliament by
the Secretary of State for Health
by Command of Her Majesty
December 2009



Government Response to the
House of Lords Science and Technology
Committee Inquiry into
Genomic Medicine

Presented to Parliament by
the Secretary of State for Health
by Command of Her Majesty
December 2009

© Crown Copyright 2009

The text in this document (excluding the Royal Arms and other departmental or agency logos) may be reproduced free of charge in any format or medium providing it is reproduced accurately and not used in a misleading context. The material must be acknowledged as Crown copyright and the title of the document specified.

Where we have identified any third party copyright material you will need to obtain permission from the copyright holders concerned.

For any other use of this material please contact the Office of Public Sector Information, Information Policy Team, Kew, Richmond, Surrey TW9 4DU or e-mail: licensing@opsi.gsi.gov.uk.

ISBN: 9780101775724

Printed in the UK by The Stationery Office Limited
on behalf of the Controller of Her Majesty's Stationery Office

ID P002338539 12/09 958 19585

Printed on paper containing 75% recycled fibre content minimum.

Introduction

This document sets out the Government's response to the House of Lords Science and Technology Committee report on Genomic Medicine, chaired by Lord Patel. Detailed responses to each of the 54 recommendations contained in the Committee's report can be found from page 7 onwards.

Ever since Watson and Crick discovered the double helix structure of DNA in 1953, the UK has led the world in genetics research and genetics based healthcare, pioneering major achievements in the field such as DNA fingerprinting, chip technology and gene sequencing techniques. This includes the major role the UK played in the completion of the Human Genome Project (HGP). This revolutionary work is still providing new avenues for research into understanding and improving our health and wellbeing.

The Government remains committed to genetics research and aims to maintain the UK's position as a world leader in associated health research, development and innovation.

As we advance our knowledge of human genetics, this will continue to play an increasingly important role in our healthcare. With the founding principle of universal healthcare for all, available free at the point of delivery, the NHS is well suited to adopt the benefits that genetic discoveries bring. This founding principle has protected, and will continue to protect, the public against discrimination on the basis of their genetic information.

The Select Committee's report on the rapid developments in human genetics is extremely timely and builds on previous work by their predecessors in the Lords Select Committee on Science and Technology Report on Human Genetic Databases published in March 2001. That report identified the potential benefits of genetic research in understanding complex diseases in healthcare and in pharmacogenetics. The Government took careful note of many of the recommendations in developing the Genetics White Paper 2003 *Our Inheritance, Our Future: Realising the Potential of Genetics in the NHS* (the White Paper). The White Paper had as its foundation the UK's reputation as one of the pioneers of genetic research, with a leading role in the international HGP and with a well developed research and clinical infrastructure to apply research findings to medicine.

The White Paper set out a strategy for research into the link between genes and disease and to prepare the NHS to maximise use of the new knowledge. Consequently, more than £80 million of new expenditure has been allocated to fund a broad-based programme of work that has put in place a framework for NHS genetics services. The programme was reviewed in 2008 and several initiatives were refreshed, including strengthening of specialised genetics services, building genetics into mainstream services, spreading knowledge across the NHS and generating new knowledge and applications.

The Government is pleased that the Committee recognised the considerable achievements that have been made since the publication of the White Paper. We share the Committee's view that the pace of development in genome sequencing, advanced genetic diagnostics and understanding of the linkages between genetic variations and diseases has exceeded our expectations. The Government believes that many of the initiatives established at the time of the previous report and White Paper have contributed to this rapid pace of development.

The research funders, led by the Medical Research Council (MRC) have invested in infrastructure, specific project funding and capacity building. Collectively, this amounted an investment of more than £45 million in genomic medicine in 2007-08.

In 2009, more than £9 million was awarded by the MRC to support the UK research community's access to high quality equipment for DNA sequencing via substantial investment in the latest technology. Four regional hubs located across England and Scotland will provide technical support and bioinformatics expertise. These hubs will allow academics to capitalise on high throughput, DNA sequencing machines and expand the potential of these resources. For example, the Eastern Sequencing and Informatics Hub (EASIH) in Cambridge will provide researchers with access to the tools required to analyse complicated datasets. The hub, in collaboration with the National Blood Service for England and Wales and NHS Diagnostic Services, will use high throughput sequencing for routine medical diagnostics, including tissue typing for transplantation, cord blood stem cell transplants and prenatal diagnosis. The Department of Health (DH) also funds work through the National Genetics Reference Laboratory (NGRL) Wessex to help co-ordinate successful adaptations of Next Generation Sequencing for targeted diagnostic use.

While a great deal of basic research is still needed to understand the complex insights we now have, it is clear that the ability to stratify groups of patients in new ways will change how we discover, evaluate and use future medical treatments. The ability to develop sophisticated diagnostic approaches, and use them well, will be central to achieving better outcomes. The MRC, as the lead for the Office for the Strategic Co-ordination of Health Research (OSCHR), is developing a stratified medicine strategy in partnership with the Technology Strategy Board (TSB). The TSB and MRC have organised a stakeholder workshop that will inform the MRC's strategy and investment plans and the case for a potential innovation platform on stratified medicine. In parallel, the MRC held a joint workshop with the Association of the British Pharmaceutical Industry (ABPI) on immunology and inflammation in July 2009. Patient stratification was a key element of this workshop that involved leading academics and representatives from major healthcare companies. Following from the first workshop, the MRC will develop a research agenda around the priorities identified, focusing initially on specific disease areas.

The Economic and Social Research Council (ESRC) is providing significant contributions to research on the economic and social aspects of genomic medicine and in 2008-09 will spend in excess of £5 million on these topics; this will include investment in the ESRC Genomics Network (EGN). One of the four research themes of the EGN is 'biomedicine, health and identity'. This research covers the social, economic, ethical, political and legal aspects and implications of genomics-related developments, including the application of genomics in healthcare settings.

The TSB is acting as a catalyst for business innovation with a view to enabling the development of the next generation of technologies in disease prevention, diagnosis and treatment through funding collaborative research and development projects between academia and industry. The DH, with the Department for Business, Innovation and Skills (BIS; at the time it was the Department of Trade and Industry), funded the genetic knowledge parks and, subsequently, broadened these to support the biomedical research centres. All of these initiatives have increased the knowledge of the links between genetic changes and diseases.

The MRC supports large patient cohort studies that have been established to investigate the links between genetics and a range of common diseases and conditions. This support continues. For example, in 2009, funding was awarded for research on the stratification of treatment for children (aged 1-5 years) with pre-school wheeze. The aim of the research is to determine, through the measurement of genetic variants, whether there is a subgroup of children who are highly responsive to the oral medicine montelukast (Singulair®). The study will be the largest trial currently conducted in wheezy pre-school children and may open up genetic testing for the condition. If this large study shows an overall beneficial effect of intermittent montelukast therapy, this approach will be integrated into the British Guidelines on the Management of Asthma, and is potentially applicable to 10-20 per cent of the pre-school population. Even a modest effect on unscheduled healthcare attendance, at the individual level, will translate into a significant impact on health, since the prevalence of the condition is very high in the UK.

The advances in genetic knowledge will undoubtedly affect virtually all aspects of medicine. Genetic and genomic techniques can be used to:

- Predict the onset or severity of diseases;
- Identify sub-types of common diseases;
- Find new targets for novel medicines; and
- Help predict the response to medicines.

However, the knowledge of such genetic links does not necessarily lead to the availability of diagnostic tests or therapies. There is a need to evaluate the accuracy and predictivity of genetic markers to inform their clinical use. The NHS currently offers more than 400 genetic tests for single gene disorders through the

world-leading UK Genetic Testing Network (UKGTN). The UKGTN has developed methods to evaluate new tests and to ensure that the NHS laboratories offering tests do so accurately and efficiently. DH has invested over £18 million on new laboratory equipment for NHS genetic laboratories and this initial funding is now being built upon through investment from the NHS. Further investment is being made by NHS Trusts and Strategic Health Authorities (SHAs) as the technology develops. The Government's aim is to ensure that high quality and equitable genetic services are accessible throughout the NHS.

We concur with the Select Committee's opinion that education is key to the mainstreaming of NHS genetics services. We have funded the NHS National Genetics Education and Development Centre (NGEDC) to develop a continuum of genetics education, identifying learning outcomes for pre- and post-registration education and genetics activities for the workplace. This will support the clinical application of genetics knowledge, skills and activities for non-genetics healthcare workers. These elements of genetics are common core components, able to be adapted for use in the education of different health disciplines and roles.

We believe these programmes have been successful because their delivery is based firmly in the NHS. There has been widespread collaboration and consultation with providers of education, commissioners of services, the genetics services and groups of health professionals and their trainers.

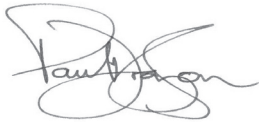
As well as education, we have also had measurable success in developing the 'right staff with the right skills' to provide NHS genetics services. The White Paper made a commitment to fund up to 90 Grade A trainees in laboratory genetics. By the end of the 2012-13 financial year we will have invested approximately £15 million in this scheme. This will ensure the provision of more than 90 Grade A trainees. This is not the only training initiatives we have instigated in our efforts to provide genetics services within the mainstream NHS: programmes are in place to provide more than 50 genetic counsellors, at a cost of £3.2 million. The final part of this process is now underway in partnership with NHS Trusts through a two-year training programme which will be completed in 2010-12. In addition, as part of the Modernising Scientific Careers programme, which began in October 2009, DH is investing over £4.5 million to address the training needs of healthcare scientists in genetics laboratories.

Throughout the report from the Select Committee, the term 'genomic medicine' is used. The Government acknowledges that the Committee seeks to make a distinction between traditional genetic techniques and the newer advances in genomic science. However, to ensure clarity, this response will refer in places to the existing terms genetics and clinical genetics, as these terms are most readily understood and align most closely with existing professional disciplines. We accept that clinical geneticists and other clinical specialties are now using genomic techniques. The Government has promoted this through a number of White Paper initiatives under the heading 'mainstreaming genetics'. Whilst we may not use the same terminology, the Government agrees with the Select Committee that gene based knowledge will play a key role in the future

provision of healthcare across the NHS. The extent of that role, however, still needs to be determined fully, and the Government hopes that this response is clear in establishing how we will achieve that aim. We need to ensure that the NHS is ready for future developments and that new technologies are properly introduced, without hindrance, from laboratory bench to bedside.



Gillian Merron, Minister of State for Public Health



Lord Drayson, Minister of State, Department for Business, Innovation and Skills

Government Response to the House of Lords Science and Technology Committee Inquiry into Genomic Medicine

- 1. We recommend that OSCHR should take the lead in developing a strategic vision for genomic medicine in the UK with a view to ensuring the effective translation of basic and clinical genomic research into clinical practice. This strategic vision should form the basis of a new Government White Paper on genomic medicine (Paragraph 8.2).**

As the Select Committee has acknowledged, the investment made to date through the White Paper has already provided new levels of genetic knowledge, skills and services within the NHS. It set out a strategy for research into the link between genes and disease and to help the NHS to maximise use of this new technology for patient benefit. The White Paper was reviewed in 2008 and, as a result, DH is taking several actions. These actions include the strengthening of specialised genetics services; positioning genetic services into the mainstream diagnostic pathway; promoting understanding across the NHS; and generating new knowledge and applications.

The White Paper and its consequent review are proof of the Government's commitment to a strategic vision to use genetic and genomic advances in the UK. It is important that any strategy for future NHS clinical services builds upon the extensive work that has already been conducted via this initiative.

The Select Committee's report, as we indicate below, identifies a wide range of matters that will need to be addressed if the benefits of genomics for medicine are to be realised. These go beyond the important role of OSCHR to facilitate more efficient translation of health research into health and economic benefits in the UK, through better co-ordination of health research and more coherent funding arrangements to support that translation. This has been successful and resulted in an increased focus on translational research to fill the major gaps in public funding and in building capacity. We consider there is a need for cross-Government action to develop strategic vision for genomics in the NHS further. DH, we believe, is best placed to take this forward as a continuum to the White Paper implementation, working in close partnership with BIS, the Research Councils, the National Institute for Health Research (NIHR), the TSB and other stakeholders, with OSCHR playing a key role in this development. As part of this process, we will invite OSCHR to consider developing a strategic vision for genomic translational research.

We also recognise that any future strategic vision must clearly show how each step of the journey from bench to bedside is linked and shares the common purpose of improved health services. As a way of achieving this objective, we will establish a cross-departmental Human Genomics Strategy Group (HGSG), which will comprise of key individuals and organisations in the field of genetic research and its application to medicine. HGSG will monitor advances in genetic and genomics research, both basic and translational, to evaluate their benefit to healthcare services in the NHS. This will enable the continued progression of the White Paper initiatives, whilst harnessing the potential of advances made in research and technology in the field of genomics. In partnership with other stakeholders, HGSG will develop a vision for genomics in the NHS. This framework, informed by the findings contained in its committee's report, will form a basis for the continued development of all aspects of genomic medicine and its integration into NHS health services. HGSG will report annually on its progress.

HGSG will provide findings to other relevant committees (including those that consider new medical treatments and procedures), reporting on their impact on services and how they might be introduced into mainstream practice. This would include, for example, the Ministerial Industry Strategy Group (MISG) and the Ministerial Medical Technology Strategy Group (MMTSG). The remits of MISG and MMTSG are to facilitate engagement between the government and the two, key supply industries (biopharmaceutical and medical technology) on important issues such as innovation in medicines and medical devices and diagnostic technologies.

In view of this, the Government does not believe there is a need for a new White Paper on genomic medicine at this time. Many of the initiatives from the 2003 White Paper are still being implemented. Therefore, we believe that the application of genomics in the NHS will be better served if we continue to build upon the excellent work that has already been conducted.

2. We recommend that the Government revises the UK implementation of the EU Clinical Trials Directive, in consultation with the research community, to make it less burdensome for researchers (Paragraph 8.3).

If the European Commission decides in favour of a review of the EU Clinical Trials Directive in 2010, we urge the Government to participate fully in discussions in order to ensure that the revised Directive is less burdensome for researchers (Paragraph 8.4).

The Government is committed to embedding the principles of proportionate, risk-based regulation across all regulated sectors. This includes cutting bureaucracy and unnecessary 'red tape' to deliver greater accountability and better focused, better targeted and more effective protections.

DH, the Medicines and Healthcare Products Regulatory Agency (MHRA) and the MRC are currently considering what changes need to be made to the EU Clinical Trials Directive to ensure the regulatory framework is fit for purpose. The MHRA is also working with their EU counterparts to identify any issues at a European level. Should the Commission decide to review the EU Clinical Trials Directive, the Government will participate fully in discussions with other member states on potential changes to the Directive through its representatives on the relevant European committees.

3. We recommend that the proposed White Paper on genomic medicine (see Recommendation 2) and the Strategic Vision of the Office for the Strategic Co-ordination of Health Research should identify barriers to collaborative working between academia and the pharmaceutical and biotechnology industries, and ways of removing them and also address the need for incentives for collaboration so as to promote translational research in the UK (Paragraph 8.5).

As stated previously, we do not accept the case for a new White Paper on genomic medicine at this time. However, we do agree that any barriers to collaborative working need to be identified and addressed and this will be part of the remit of the HGSG. Considerable work is already being conducted through the implementation of recommendations made in Sir David Cooksey's *A Review of UK Health Research Funding*, published in December 2006. For example, the TSB is continuously looking for ways to improve collaborative working using its collaborative research and development funding mechanism, Knowledge Transfer Networks (KTNs) and partnerships and innovation platforms. This helps the development of consortia and collaborative groups and creates 'innovation supply chains'. Such innovations are developed in partnership with the Research Councils, regional development agencies and the devolved administrations to address many of the challenges experienced in collaborative working.

The *Life Sciences Blueprint*, published in July 2009, also identified and addressed barriers in collaborative working, proposing a new Research Excellence Framework (REF) to replace the Research Assessment Exercise (RAE) from 2010. As set out in the *Life Sciences Blueprint*, the new REF will explicitly assess the economic and social impact of research, taking into account, for example, the translation of research into new products and services, and collaborative working between academia and business, and between academia and public services and policy makers. The new REF will be announced in 2010.

Following the launch of the Life Sciences Blueprint, OSCHR is already leading on the establishment of a series of Therapeutic Capability Clusters, the initiative by which industry, academia and the NHS will focus on areas of translational medicine, particularly early and exploratory development, where the potential for collaboration is substantial.

4. We recommend that the National Institute for Health Research ring-fence funding, through a specific Health Technology Assessment programme, for research into the clinical utility and validity of genetic and genomic tests within the NHS (Paragraph 8.6).

We recommend that the Department of Health extends the remit of the National Institute for Health and Clinical Excellence to include a programme for evaluating the validity, utility and cost-benefits of all new genomic tests for common diseases, including pharmacogenetic tests (Paragraph 8.7).

The Government agrees with the Committee: we recognise the importance of correctly evaluating and validating genetic tests. The NIHR fully allocates its funding resource to the research programmes and activities detailed in *Best Research for Best Health: A New National Health Research Strategy*, published in 2006, and these programmes provide the necessary framework to evaluate genetic and/or genomic tests. As Professor Dame Sally Davies, Director General of Research and Development and Chief Scientific Adviser to the NIHR, explained to the Committee when giving evidence in January 2009, DH is already putting more money into the Health Technology Assessment (HTA) programme. Any appropriate research projects can apply for funding, including those concerned with the clinical utility and validity of genetic and genomic tests in the NHS. We do not agree, therefore, with the recommendation to 'ring-fence' funding through a specific HTA for research into the clinical utility and validity of genetic and genomic tests within the NHS.

DH recognises the crucial role that diagnostics play in the prediction of certain diseases. As part of its programme to encourage faster uptake of beneficial new medical technologies by the NHS, DH has commissioned the National Institute for Health and Clinical Excellence (NICE) to develop and manage a single evaluation pathway, specifically for medical and diagnostic technologies. NICE is a world leader in the assessment of new medicines and has produced guidance on a number of diagnostic tests and procedures through appraisals, interventional procedures and clinical guidelines programmes. The programmes will focus on those diagnostics likely to benefit from national evaluation and have a significant positive impact on the delivery of health services in England. The criteria will be agreed in consultation with the full range of relevant stakeholders including manufacturers, clinicians and academics. Genetic tests defined as *in vitro* diagnostic (IVD) medical devices under the European IVD Medical Devices Directive are expected to fall within the scope of the programme provided they have a medical, as opposed to lifestyle, purpose.

Activity is already underway in the form of a pilot scheme and will continue into the early part of 2010. The pilot scheme will also shed light on what capacity is needed – the nature of this work is developmental and the intention is not to assess all new diagnostics as they emerge but to focus on those best suited to this type of approach. Initially, therefore, capacity will be limited and the pilot scheme will provide valuable information for decisions on future capacity and the appropriateness of methods used. Work on the diagnostic programme is at an early stage and details concerning development of methods and how to take account of clinical utility are still being considered. The first outputs of the pilot scheme are likely to emerge in summer 2010. The programme will produce guidance for the NHS on the efficacy and cost-effectiveness of diagnostic tests.

A new committee will also be established within NICE to consider proposals for the new diagnostic programme. Selection of the membership is to occur early in 2010 and the initial meeting to induct appointees will take place in the Spring. The committee's first task will be to look at the outcomes of the diagnostics pilot activity.

Furthermore, the UKGTN, which is funded by the DH and located in the National Specialised Commissioning team in London NHS, already advises the NHS on genetic testing for inherited disorders. This involves evaluating new diagnostic tests and making recommendations to commissioners on new NHS services. The UKGTN also provides an advisory role to DH on national policies.

- 5. We recommend that the Government support the re-classification of genetic tests to “medium risk” in the current review of the EU In Vitro Diagnostic Medical Devices Directive so as to ensure that all genomic tests on the market have been subject to pre-market review before their use either by the consumer directly or by the NHS and private healthcare services (Paragraph 8.8).**

The Minister for Public Health accepted in her evidence to the Select Committee that the UK would support a global harmonisation task force model for the higher classification of genetic tests. In response to the EU consultation on proposals for a recast of the Medical Devices Directive (MDD), the UK called for the European Union IVD MDD to be reviewed. The European Commission has already established a technical working group to consider any changes and we continue to press for re-classification through our representatives from the MHRA. However, the Government wants to ensure that any such changes are brought about in consultation with all stakeholders and do not place unnecessary burden on the NHS or industry.

- 6. We recommend that the Government continue to work with the pharmaceutical industry to extend value-based pricing for the stratified use of medicines under the PPRS to reflect the value of drugs sold for stratified use and the increasing use of genetic tests to accompany such treatments (Paragraph 8.9).**

We recommend further that, with regard to medicines for common diseases which are already in use in the NHS, the National Institute for Health Research should target funding to encourage the development of pharmacogenetic tests to stratify use of these medicines in order to improve their efficacy and to reduce the frequency of adverse reactions (Paragraph 8.10).

We recommend that the Department for Innovation, Universities and Skills address the issues relating to the management of intellectual property rights within the healthcare sector to improve incentives for stratifying uses of new and existing medicines and for development of pharmacogenetic tests necessary for stratification (Paragraph 8.11).

Following the challenge set by the Office of Fair Trading report on the Pharmaceutical Price Regulation Scheme (PPRS), which recommended that the Government ensure that drug prices better reflect their therapeutic value, the Government and industry recognised there was scope to improve the way in which drug prices reflected value. The PPRS has, therefore, recently been renegotiated to ensure better value through new and more flexible pricing arrangements and a more systematic approach to the use of patient access schemes. The new PPRS will be capable of better reflecting value for stratified medicines, and for other medicines. Both patient access schemes and flexible pricing may be relevant and are potentially available as options for any drug that is part of a NICE appraisal.

The PPRS is a five-year agreement between the Government and the ABPI. A new PPRS cannot be brought into operation before 2014 and it is not possible to amend the current agreement. However, patient access schemes and flexible pricing will be reviewed during the course of the current PPRS. The points raised in this report have been noted and, if there are specific issues that arise in operating the PPRS, these issues will be addressed in the next renegotiation or in the planned review of flexible pricing and patient access schemes, as appropriate.

Prior to renegotiation of the PPRS, value was already a factor in the way drugs were purchased by the NHS through encouraging manufacturers to set prices that reflect value and to support uptake by the NHS. Also, the NICE appraisal guidance clearly indicates to the NHS whether and under what circumstances the use of a particular drug is both clinically appropriate and cost-effective. NICE has already issued appraisal guidance on some drugs for stratified use, including the cancer drugs trastuzumab (Herceptin®) and cetuximab (Erbix®).

Having the right framework for the management of intellectual property is critical to ensuring that good ideas and inventions that arise within the broad variety of NHS staff – nurses, doctors and researchers – are developed to their full extent, to bring benefits to the NHS and its patients. The NHS also depends on having access to genetic tools to develop diagnostics and these may be dependent on obtaining licences to intellectual property held by companies. In line with the public sector innovation and procurement agenda, there may also be ways to make the licensing-in of genetic technologies more effective. Establishing a cohesive policy to achieve this complex set of objectives presents significant challenges. Without thorough consideration of these issues there is a risk of adverse consequences in the delivery of effective diagnostics, staff morale and healthcare expenditure.

BIS, which is responsible for intellectual property, is working closely with DH to ensure that the intellectual property system and the management of intellectual property rights are supportive of the national strategy on stratified medicine.

7. We recommend that the Department of Health set out a national strategy on stratified uses of medicines (as part of the proposed White Paper on genomic medicine (see Recommendation 2 above). The purpose underlying this strategy should be to streamline the co-development of stratified uses of medicines and of pharmacogenetic (or other) tests (Paragraph 8.12).

The Government's response to the Review and Refresh of Bioscience 2015 report (BIGTR2) in May 2009 outlined the Government's commitment to the development of a stratified disease strategy as a priority area for the TSB and MRC. The TSB and MRC, under the auspices of OSCHR, are working together to help co-ordinate the activities of public sector organisations, including the regulatory authorities and government departments, to encourage the development of the optimal research, regulatory and fiscal environment in which stratified approaches to healthcare can flourish. The importance of the co-development of stratified uses of medicines and of pharmacogenetic tests is a key part of this work. Recognising the opportunities as well as the challenges presented by stratified medicine, the TSB and MRC are also working in partnership to explore the case for developing a new innovation platform in stratified medicine. An update on work in this area will be included in the BIS co-ordinated report to ministers (to be published in January 2010) on progress against the BIGTR2.

The importance of pharmacogenetic testing is already established. As part of the commitments in the White Paper, DH has appointed Professor Munir Pirmohamed as the first NHS Chair of Pharmacogenetics, located at the University of Liverpool, in the new Wolfson Centre for Personalised Medicine. The University's Department of Pharmacology is recognised worldwide as a leader in developing the area of stratified medicine. The aim of the department is to create a centre of excellence in advancing patient treatment in the NHS. Current models of treatment rely on a 'one dose fits all' approach but, with the recent advances in genomics, there is the possibility we will be able to refine treatment so that patients get 'the right medicine, at the right dose' to maximise efficacy and minimise toxicity. An ultimate aim of the Wolfson Centre for Personalised Medicine will be to work with NHS partners to allow implementation of genetic tests into NHS clinical practice in a timely and cost-effective manner. This is the first centre of its kind in the UK.

In its 2008 GO-Science Review of the Department of Health, the Government Office for Science stated, 'The strategic approach used by DH to support pharmacogenetics is a model that might be considered for other areas of new or under-resourced science.' It went on to say that DH had provided targeted support to the area of pharmacogenetics research that focused on existing medicines that patients are currently taking, and which is unlikely to be addressed without public sector funding (i.e. addressing a market failure). This was considered as an example of targeted funding at an identified research need, with significant potential benefits in terms of improved patient health and reduced NHS costs.

In October 2009, a forum on personalised medicine was jointly organised by the MHRA and the ABPI to explore issues arising from the development of personalised medicines. Personalised medicine is a very broad topic, and the scope of the forum discussions included not just issues associated with drug dose but also drug choice and targeting drugs to specific patient groups. The event brought together experts representing a wide range of interests relevant to this issue, including academics, industry, regulators and patient/lay representatives. Professor Munir Pirmohamed chaired the event. A report will be published by the MHRA and may be used to make proposals for future developments on this issue.

Key issues raised during the forum, which are thought to warrant further consideration by the MHRA and ABPI, included: the importance of ensuring that the regulatory frameworks can accommodate the parallel development of diagnostics and drugs targeted at specific patient populations; the impact that targeted medicines will have on current clinical trial design; and the mechanisms needed to ensure that, as diagnostic tests are developed and refined for targeting existing medicines, they can be subject to appropriate regulatory control. The importance of gaining clinician recognition of the need to resource and use diagnostics is also a key issue for the future.

As the area of stratified medicine is developed, the Government will work to ensure that the NHS harnesses any benefit in an effective and efficient manner. The measures referred to above, combined with our plans for a HGSG, place the UK in a prime position to ensure this is brought about.

8. We recommend that genomic science is adopted as a key technology platform by the Technology Strategy Board, to drive forward commercial development and clinical application in this area over the next five years and to maintain the UK lead in genomic medicine (Paragraph 8.13).

The TSB recognises the importance of genomics and genomic technologies and their application into human healthcare and as a growing business opportunity. This is reflected in two TSB strategies in the bioscience technology and the medicines and healthcare application areas. Genomics is one of three technology pillars in the TSB biosciences strategy highlighting the opportunities for application in predictive biology (including systems approaches to predictive safety testing), understanding the genetic basis of disease and the development of new diagnostic tests.

The medicines and healthcare strategy addresses genetic factors in the stratification of patients and its role in providing effective treatment options and for developing preventative strategies, particularly in the area of chronic disease. Stratified medicine is an increasing challenge for healthcare providers and represents an opportunity for the UK's pharmaceutical, diagnostics and devices sectors. TSB is currently working in partnership with the MRC to explore the case for developing a new innovation platform in stratified medicine; this could lead to up to £50 million of new investment in the area beginning in 2010-11. Other options to promote collaboration, engagement and networking between companies, academics and clinicians particularly in the diagnostics sector, will be taken forward through the TSB's new health technologies and medicines KTN.

9. We recommend that the Government should reconsider how they will prepare NHS commissioners and providers for the uptake of genomic medicine in the NHS. We also recommend that the National Institute for Health Research, as part of its remit, regularly monitors developments in genomic medicine and their implications for the NHS now and in the future (Paragraph 8.14).

We envisage that the proposed White Paper (see Recommendation 2 above) will address the operational changes needed as a result of bringing genetic aspects of treatments for common disorders into mainstream clinical specialities (including changes to commissioning arrangements, processes for providing genetic tests within the NHS and arrangements for NHS laboratories to conduct such tests) (Paragraph 8.15).

We recommend that, on the basis of the monitoring activity of the National Institute for Health Research (see Recommendation 14 above), the Secretary of State for Health should ensure that any necessary NHS operational changes, as a result of a shift in the provision of genomic services to mainstream medicine in the NHS are implemented in the NHS. In order to facilitate the process the Secretary of State should identify whether the NHS is fit to handle such changes and also what new service models are needed if health professionals from other clinical specialties are to take routine responsibility for genomic aspects of healthcare (with referral to specialist genetics services only where necessary) (Paragraph 8.16).

We recommend that the Department of Health should conduct a review with the aim of establishing appropriate commissioning structures for pharmacogenetic tests, tests for management of genetically complex diseases and tests for diagnosing single-gene subtypes of common diseases, as the use of such tests spreads further into the mainstream NHS (Paragraph 8.17).

We recommend that the Department of Health should conduct a review of current genetic test service provision within the NHS both for single-gene disorders and for single-gene subtypes of common disorders. This should aim to eliminate what are serious inconsistencies in the provision of genetic services across the NHS (Paragraph 8.18).

We recommend that the Department of Health should develop a national set of standards and tariff guidance for the commissioning of genetic tests, taking into account the recommendations from the second phase of the *Carter Review of NHS Pathology Services* that there should be tariff guidance for community-based and specialist pathology, particularly relating to DNA and RNA-based genetic tests (Paragraph 8.19).

The NIHR commissions and funds NHS and social care research that is essential for delivering our responsibilities in public health and personal social services. Its role is to develop the research evidence to support decision making by professionals, policy makers and patients, make this evidence available, and encourage its uptake and use. As the NIHR funds research, not implementation or service development, its contribution and advice in the area of disease prevention and treatment is always welcome. However, we do not believe that the NIHR is best placed to prepare commissioners and providers for the uptake of genomic medicine in the NHS.

It is for other organisations, such as NICE, to provide national guidance on the prevention and treatment of ill health. For example, NHS clinicians will be supported through NHS Evidence. Launched in April 2009, NHS Evidence is a world-leading online web portal, managed by NICE, that empowers staff with the world's best evidence and best practice information. NHS Evidence allows people working across health and social care to access a comprehensive range of clinical and non-clinical evidence to help them make informed decisions about treatments and resources. The new system is built around a powerful search engine that consolidates information from a wide range of sources including clinical, commissioning, drugs and technology, public health, social care and education.

Users are able to upload and share their own content (such as local service models and policies) and customise the service based on their own preferences: for example, to access evidence tailored to their needs and to receive alerts about new information. In addition, NHS Evidence will identify the best evidence by sorting, sifting and prioritising a range of information and awarding an accreditation mark to the most reliable and trustworthy sources. All information submitted for accreditation will be assessed by an independent advisory committee managed by NICE.

When discussing improvements in the commissioning of services, it is important to distinguish between the possible two possible definitions of commissioning in this context. First, the term is used in reference to the process of ordering and use of genetic testing services. Second, there is the work that DH is leading to promote the benefits of the World Class Commissioning (WCC) programme when providing NHS health services.

The Government believes that the genetic analysis of common complex diseases will only gradually become a part of NHS service provision. As such tests are required, it will be possible to include these diagnostic tests within current arrangements. Pharmacogenetic testing would follow the same commissioning processes and mechanisms that are currently used when prescribing medicines for NHS patients. DH considers that suitable commissioning structures are in place but will continue, via the UKGTN, to monitor commissioning structures within genetics and genomics.

The UKGTN is currently undertaking a review of service provision within the NHS both for single gene disorders and for single gene subtypes of common disorders. The Specialised Commissioning Group Directors' Network is also completing documentation on the designation of providers for clinical genetics. This was a recommendation from Professor Sir David Carter in his *Review of Commissioning Arrangements of Specialised Services*, published in May 2006 with the aim of addressing inconsistencies in the provision of genetic services across the NHS.

The UKGTN is also working with specialised commissioners of genetics services to determine the models and mechanisms for commissioning in each area to inform a consistent approach. This work is one of the current steps the DH is taking to embed and mainstream genetic services within the NHS. It supports the work of the Genetics Commissioning Advisory Group (GenCAG), established under the White Paper, with the remit to take a strategic national overview of genetics in healthcare delivery. It aims to provide advice to commissioners of genetics services to enable them to provide appropriate services for NHS patients and their families.

The development of national standards and tariff guidance for the commissioning of genetic tests is worthy of consideration. However, before any such guidance could be considered, it is essential that the possible impact on current service provision is analysed. This would include ensuring that any tariff, in providing economies of scale, would not do so at the expense of higher costs for tests for more rare genetic conditions. DH will discuss this matter further with commissioners before coming to a decision on the way forward.

The WCC programme supports Primary Care Trusts (PCTs) to improve health outcomes and reduce inequalities by providing evidence-based, high-quality services, offering patients choice and control and ensuring better value for all.

WCC sets out 11 competencies against which PCTs commissioning capabilities are measured and then assessed through an annual assurance process. A number of these are relevant to how PCTs consider the role of genomic medicine in the delivery of services. In particular:

- **Competency 4** – the role of clinicians and the dissemination of information to support clinical decision-making;
- **Competency 7** – stimulation of the provider market through a knowledge of current and future capacity plus capability and creation of effective choices for the patient; and
- **Competency 8** – the promotion of improvement and innovation.

As part of this process, PCTs should be prepared for ongoing changes in service delivery and scientific development, including the uptake of genomic medicine in the NHS when thinking and planning strategically to meet the needs of local populations in the most effective and efficient way. As such, WCC is not intended to be prescriptive about the types of services commissioned or where they are commissioned.

The Government recognises that the depth and breadth of the work already underway on the provision of genetic services commissioning, its structures and its delivery, is considerable. As part of our work to mainstream these services into the NHS diagnostic pathway, the Government will ask the HGSG to consider how genetic services commissioning in the NHS can be further improved. DH would not want to see any departure from this flexible approach to commissioning and will continue to concentrate on the mainstreaming of pharmacogenetic and other genetic testing into the diagnostic pathway. We will, however, ask the HGSG to monitor developments in genomic medicine and their current and future implications for the NHS.

10. We recommend that the Department of Health should commission the National Institute for Health and Clinical Excellence to issue guidance on the use of genetic tests by non-genetic specialties; and that the NHS should consider the expansion of the “red flag system” to alert healthcare workers to the need to conduct a specific test, in some cases a pharmacogenetic test, before deciding on treatment or prescription (Paragraph 8.20).

The UKGTN already provides guidance on the use of genetic tests for single gene disorders and tests for single gene sub-types of common disease for both genetics specialties and non-genetic specialties. This guidance is in the form of testing criteria available on the UKGTN website. The UKGTN also provides advice for genetic tests commissioned outside of clinical genetics. We therefore do not believe there is a need for NICE to issue guidance.

The NGEDC provides a range of initiatives and educational tools for the use of all NHS staff. As well as extensive online support, the NGEDC also provides teaching packages aimed at raising awareness of the benefits of using genetic information in diagnostics.

DH’s Genetics Information Technology (IT) Development Group aims to bring together policy leads on genetics and pathology with NHS Connecting for Health (CfH) and the NHS Information Centre to develop a co-ordinated approach to the development of IT in the field of genetics. The group is already considering how genetic informatics can be an integrated part of the National Programme for IT (NPFIT) in the NHS.

11. We recommend that the Government centralise laboratory services for molecular pathology, including genetic testing, in line with the recommendations of the second phase of the Carter Review of NHS Pathology Services. The aim should be to organise effective laboratory services for molecular pathology and genetics by bringing together the whole range of DNA and RNA-based tests for pathology and medical specialties to ensure that services are cost effective. This would have the potential to free up funds, for example, for the highly specialised technical equipment that is needed (Paragraph 8.21).

The Government supports, in principle, the approach of bringing together molecular pathology and genetics laboratories. This has the potential to benefit patients through better use of the laboratory workforce and more effective uptake and use of new molecular technologies and equipment. DH has asked SHA Medical Directors to lead work on pathology service redesign in their localities. DH will also ask them to consider how they might bring relevant pathology and genetics laboratory stakeholders together locally to consider this recommendation in light of current and future local service provision and needs.

12. We recommend that the Government show leadership on leveraging sustainable funding to the European Bioinformatics Institute (EBI), through the European Research Infrastructure (ESFRI) instrument and through the UK Research Councils. This would reduce the dependence of the EBI on charitable and cyclical funding and allow further growth of the Institute commensurate with the recent growth in genomic databases and the value of the EBI to the UK science base (Paragraph 8.22).

The UK is leading discussions at a pan-European level to help develop a more secure funding structure for the EBI. Since 2008, Research Councils UK (RCUK) has made it a priority to provide capital expenditure to renew computing facilities at the European Molecular Biology Laboratory – European Bioinformatics Institute (EMBL-EBI). This commitment has been reiterated in the draft 2010 RCUK Large Facilities Roadmap. This forms a key part of the emerging pan-European science project, the European Life Science Infrastructure for Biological Information (ELIXIR), an initiative involving 32 partners from 13 countries aimed at establishing an infrastructure for biological information in Europe that attracts sustainable funding.

The expansion in EMBL-EBI I data management capacity is vital in underpinning the sustainable development of the substantial investments in genetic, genomic and systems biology made by the Research Councils. The UK's involvement in ELIXIR is supported by the Biotechnology and Biological Sciences Research Council (BBSRC), the Natural Environment Research Council (NERC), the MRC and the Wellcome Trust. As part of this support, in August 2009 the BBSRC made available £10 million to EMBL-EBI for work to increase the institute's data storage and handling capacity. The business case for longer-term increases in the institute's data capacity is being developed.

13. We recommend the establishment of a new Institute of Biomedical Informatics to address the challenges of handling the linking of medical and genetic information in order to maximize the value of these two unique sources of information. Such an institute would bridge the knowledge, culture and communications gap that currently exists between the expertise in NHS IT systems and bioinformaticians working on genome research. The Institute would guide the NHS in the creation of NHS informatics platforms that will interface with databases containing personal genetic data and with publicly available genome databases (Paragraph 8.23).

We recommend that the Department of Health should establish a centre for national training in biomedical informatics (within the Institute of Biomedical Informatics) with the aim of providing training that bridges the gap between health records information technology and genome informatics, and ensuring the delivery of an expert workforce for the NHS (8.24).

The Government will consider this recommendation carefully, in light of developments in the research by the EBI and service areas. DH set up the NGRLs under the White Paper and, amongst other tasks, NGRL Manchester has been taking forward work to establish a broad platform of resources for the diagnostic community. This work includes informatics resources, such as the Diagnostic Mutation Database (DMuDB), Universal Browser and Single Nucleotide Polymorphism (SNP) check.

NGRL Manchester also delivers bioinformatics training courses for molecular and cytogeneticists and clinical geneticists. In addition, as part of the Modernising Scientific Careers programme, which began in October 2009, DH is investing over £4.5 million to address the training needs of healthcare scientists in genetics laboratories. A pilot genetics training programme began in October 2009, with a modernised genetics curricula, combining both clinical molecular and cytogenetics disciplines and including the development of bioinformatics input. Thirty-two trainees will participate in the pilot on two programmes: Healthcare Scientist Practitioner Training and Scientist Training Programme.

The Government concurs that the linking of medical and genetic information is extremely important if we are to realise the full benefit for NHS patients. Therefore, we will ask the HGSG to make a detailed assessment of these recommendations, including the future provision of training in biomedical informatics in the NHS workforce and the role of the NGRLs, as part of its work to develop a roadmap for NHS genomic services.

14. We recommend that the Department of Health should implement a programme of modernisation of computing and information technology within the Regional Genetics Centres and laboratories, including an upgrade in computer hardware, software tools and communication bandwidth, in order to manage current needs of clinical and genome informatics in the Regional Centres (Paragraph 8.25).

Under the White Paper, DH has funded £18 million in expanding laboratory services for genetic testing; this investment funded new technology, including high throughput capacity robotics. In addition to this investment, £1 million was provided to support IT in genetic laboratories to improve internal and external handling of work and communications between laboratories.

It is important that any changes to IT systems to provide better access to genome informatics are integral to and compatible with current NHS IT systems, through the NHS NPfIT and NHS CfH. As explained above, the Genetics IT Development Group will consider how best to include genomic and genetic information within this framework.

15. We welcome the public engagement activities that have been undertaken so far. We urge the Government and others to continue them, building on the successful dialogue models developed by Sciencewise. We have some concern, however, that these activities have focused primarily on public understanding of single-gene disorders. We urge the Government and other relevant bodies to extend the scope of their public engagement activities to include more detailed consideration of the implications of genetic tests for common complex diseases (Paragraph 8.26).

We recommend in particular that the Human Genetics Commission should promote a wide-ranging debate on the ethical and social issues relating to genetic tests and gene associations for genetically complex diseases and how they contrast with genetic tests for single-gene disorders. The debate should aim to improve public understanding of genetic risk and predictive testing in common complex disorders (Paragraph 8.27).

We recommend further that the Department of Health should establish a comprehensive and regularly updated public information web site which would review the most recent science on the genetics of common diseases, to help the public to understand and interpret results of genetic tests (Paragraph 8.28).

The Government welcomes the support the Committee has expressed for the ongoing work of the Sciencewise Expert Resource Centre ERC in developing capacity and expertise in public engagement and supporting government departments to make effective use of this in the development of policy. The work of the Human Genetics Commission (HGC) continues to be extremely important in the promotion of debate on the ethical and social questions arising from genomic and genetic research and its applications. We have asked the HGC to consider how to generate demonstrably effective and informative debate around the issues raised by complex diseases.

The Commission has already made significant progress in generating debate and increasing understanding of genetic testing for complex conditions in its work on direct-to-consumer genetic testing. The Nuffield Council on Bioethics 2009 consultation *Medical Profiling and Online Medicine: The Ethics of 'Personalised' Healthcare in a Consumer Age* is due to publish its findings in spring 2010. We will ask Sciencewise ERC and the HGC to discuss with the Nuffield Council on Bioethics the findings, actions and priorities for future public engagement that are identified as a consequence of the consultation.

More general information on genetics and genomics, scientific research and its implications for individuals is already available from various online sources. This includes the HGC website, which is currently being reviewed, and we will ask the HGC to take this recommendation into consideration, in the context of the Government's view as outlined above, when considering opportunities for improvements to its current website.

However, the Government believes it is good practice for the clinician who commissions the genetic test, or who provides the genetic test result to the patient, to be the professional who fully explains the test results. As test results can be complex and results from testing for common diseases are dependent on a number of other genetic and environmental factors, interpretation requires contextual explanation from a professional on a case-by-case basis.

16. When developing the “safe havens” for research, recommended by the *Data Sharing Review* report, we encourage the Department of Health to consider adapting the approach developed by UK Biobank for ensuring the protection of personal privacy as an exemplar (Paragraph 8.29).

The *Data Sharing Review* report suggested that a statutory duty should be put on the Information Commissioner to publish (after consultation) a data sharing code of practice to remove “the fog of confusion” – which should include sector specific instructions where necessary. It also recommended that where there was a genuine case for removing or modifying an existing legal barrier to data sharing, “a new statutory fast-track procedure should be created”. We support these recommendations (Paragraph 8.30).

Further, we urge the Information Commissioner to publish a set of clear, feasible and proportionate guidelines, in accordance with the Data Protection Act 1998, specifically for researchers handling genetic data for the purposes of non-personal research in order to reduce the burden of data protection legislation on researchers (Paragraph 8.31).

The *Data Sharing Review* report recommended strongly that, due to the need for clarity over when data-sharing is appropriate under the Data Protection Act 1998, although change may be a long way off, the Government should participate “actively and constructively in current and prospective reviews of the European Directive, and assume a leadership role in promoting the reform of European data law.”. We agree. (Paragraph 8.32).

We recommend that, meanwhile, the Government should seek to amend the Data Protection Act 1998, where possible, (including amendments to bring into effect Recommendation 31 above) so as to facilitate the conduct of non-personal research using genetic data (Paragraph 8.33).

The Research Capability Programme of Connecting for Health has prepared the business case for a Health Research Support Service to provide the right environment for a number of research uses of data sets derived from patient information. These uses will include:

- Support for interventional research in which the NHS infrastructure is used to identify efficiently and comprehensively patients eligible for a specific healthcare intervention (for example, therapy or preventative activity). This will facilitate study feasibility assessments and recruitment into trials and remote data capture, hence enabling faster and cheaper clinical trials;
- Support for observational research in which data collected during the course of routine clinical care are used to study the health of the population, the natural history of disease, the safety profile and the clinical and cost effectiveness of healthcare interventions, used in daily clinical practice.

The programme includes plans to federate some valuable, large databases to create new opportunities for research. They could, in principle, include genetic information providing the research guarantees anonymity and following appropriate data protection and consent legislation.

The Government response to the *Data Sharing Review* made no specific commitments about genomics or genetic information. However, the Government is taking forward a provision in the Coroners and Justice Act 2009 to place a statutory duty on the Information Commissioner's Office (ICO) as the independent regulator of the Data Protection Act 1998 (DPA), to issue a code of practice on the sharing of personal data. The Ministry of Justice (MoJ) is liaising with the ICO on this matter. The ICO has stated that it is open to working with subject experts to produce specialist guidance. DH is currently investigating the extent of the desire to issue guidance amongst practitioners.

The Government keeps the legislative framework for data protection under constant review and is open to making amendments, should a significant body of evidence demonstrate the need to do so. This includes engaging actively in EU work on data protection.

17. We do not believe that at present there should be specific legislation against genetic discrimination, either in the workplace or generally. But rapid advances in genetic science mean that there is a continuing need to monitor the situation. This should be undertaken by a designated body, possibly the Human Genetics Commission (Paragraph 8.34).

We recommend that the Government should negotiate with the Association of British Insurers a new clause in the Code of Practice, *Moratorium and Concordat on Genetic Testing and Insurance* that prevents insurers from asking for the results of genetic tests which were carried out while the Moratorium was in place. (Paragraph 8.35).

We recommend that the Government, together with the Association of British Insurers, should establish a longer-term agreement about the use of genetic test results for insurance purposes. The moratorium is next due to be revised in 2011. This would provide a good opportunity to take this recommendation further. (Paragraph 8.36).

Given that the Genetics and Insurance Committee is to be disbanded, we recommend further that the Government should put in place arrangements for monitoring the use of genetic tests for insurance purposes. These arrangements should be part of the longer-term agreement on the use of genetic testing in insurance envisaged in Recommendation 36 above. (Paragraph 8.37).

We agree that there is no need for specific legislation against genetic discrimination at present.

The healthcare of all UK citizens, regardless of their risk, is covered by the NHS. The values of the NHS mean citizens can choose to take genetic tests free from the fear that, should they test positive, they will face an enormous bill for insurance or become priced out of cover altogether. It provides a defence against the inequality and the prospect of a 'genetic underclass' unable to access the healthcare they need.

As the Committee notes, in April 2009, the Government decided that the Genetics and Insurance Committee (GAIC) should be disbanded once alternative arrangements to cover its remaining remit were agreed.

The HGC has recently established a working group to better define genetic discrimination and to conduct a comprehensive evaluation of the risk of its occurrence. The project will include an examination of legal and philosophical concepts of genetic discrimination and the existing measures that protect against it. The project will also aim to identify any evidence of genetic discrimination and will consider appropriate monitoring and alert systems and the appropriateness of existing protections in light of anticipated developments. The Commission is planning to launch the project with a public information-gathering seminar early in 2010 and will consult widely with stakeholders and the general public throughout the project. The work is expected to conclude in 2011.

Following discussions with the relevant stakeholders, it has been agreed that the HGC will take responsibility for overseeing insurers' compliance with the *Concordat and Moratorium on Genetics and Insurance* and monitoring new developments in genetic testing and insurance. The Government agrees with the Committee that the scheduled review of the concordat and moratorium in 2011 would be the right time to examine the Select Committee's recommendations. This would inform the Government and the Association of British Insurers, when considering a longer-term agreement about the use of genetic test results for insurance purposes.

The remaining function of the GAIC was to review applications for the use of genetic test results for insurance purposes. The Government is of the view that it would be inappropriate for the HGC to undertake this function. Therefore DH will draft proposals to establish a process for convening a panel of expert and lay individuals, as required, to review any further applications from the ABI. The arrangements will be outlined in an updated version of the *Concordat and Moratorium on Genetics and Insurance*.

18. We support the Human Genetics Commission's work on developing, with the industry, a voluntary code of practice for selling genetic tests directly to consumers. The code should include a requirement for companies to place in the public domain information about the standards adhered to and the national accreditation status of the company's laboratory, and the clinical validity and utility of the tests offered. The code should also include guidelines for provision of appropriate pre- and post-test counselling and an ethical code of conduct for the sale of such tests (Paragraph 8.38).

Further to Recommendation 28 above, we recommend that the proposed Department of Health web site should set out the following:

- **up-to-date information on the national or international accreditation schemes with which the "direct to consumer" test (DCT) laboratories are registered, including the laboratories' registration status;**
- **the quality assurance schemes in which these laboratories participate; and**
- **the extent to which the DNA sequence variants used by DCTs for predicting risk of future disease have been validated in the genome-wide association studies, and shown in prospective trials to have utility for predictive genetic testing (Paragraph 8.39).**

The HGC leads an international, expert working group that has developed a *Common Framework of Principles* for DCT services. A draft of these principles was published for consultation on 8 September 2009. The principles cover all aspects of DCT services including: the marketing of tests; information that should be made available to consumers (including information about the scope of the test and the analytical and clinical validity of each genetic marker used); consent; the laboratory analysis of biological samples; and the levels of support that should accompany the provision of genetic test results. The principles are intended to address the international scope of the market in DCT and their cross-border provision and to provide a template for more specific guidance that considers existing national laws and guidance.

Adherence by providers to these principles will provide confidence to consumers that the necessary accreditation and quality assurance mechanisms are in place. The HGC is working with official bodies, industry, professional bodies and stakeholder groups internationally to promulgate the principles and to encourage adherence in their final form. Information on the *Common Framework of Principles* will be available on the HGC website (linked to the DH website).

19. We believe that understanding the use of genomic tools for diagnosis, stratification of patients and choice of treatment in common diseases should form an important part of the undergraduate medical curriculum and urge the General Medical Council to take this aspect of disease management into account in their current review of *Tomorrow's Doctors* (Paragraph 8.40).

We recommend that the Royal Colleges of Pathologists, Physicians and General Practitioners, after consultation with other relevant bodies, should develop a joint national strategy for undergraduate and postgraduate education and training in genomic medicine, with a clear timetable for implementation (Paragraph 8.41).

We recommend that the General Medical Council should introduce training in genomic medicine as a core competency in the Certificate of Completion of Training of all junior doctors training in the medical and pathological specialties (Paragraph 8.42).

We recommend that general practitioners should be trained to be able to provide general advice to patients on the implications of the results of predictive tests for common diseases. Planning how this might be done should be part of the review by the Royal Colleges recommended in Recommendation 41 above (Paragraph 8.43).

We recommend that the Postgraduate Deans of Medicine and Medical Education for England, together with the relevant Royal Colleges and the Postgraduate Medical Education and Training Board, reinstate the currently suspended training programme in genetic pathology with a view to reintroducing a viable programme for the intended small number of pathologists (perhaps up to five at any one time) training in this specialty. This training may need to be overseen by both pathologists and clinical geneticists and could lead to the possibility of dual accreditation in genetics and pathology (Paragraph 8.44).

We also recommend that the Department of Health should work with the Postgraduate Deans of Medicine and the relevant Royal Colleges to reinstate consultant posts in genetic pathology capable of absorbing a sustainable number of registrar training posts (Paragraph 8.45).

We recommend that genomic medicine is included as a clinical competency within continuing professional development (CPD) for clinicians in primary and secondary care, and that this is recognised by the Royal Colleges which monitor CPD (Paragraph 8.46).

We urge the Nursing and Midwifery Council to set detailed standards across the curriculum on genetics and genomics for nurses, both for pre-registration nursing education and as part of post-registration education and practice (Paragraph 8.47).

The Government agrees that use of genomic tools for diagnosis, stratification of patients and choice of treatment in common diseases should form an important part of the undergraduate medical curriculum. To that end, the revised version of *Tomorrow's Doctors*, which was launched on 4 September 2009, includes

a requirement that medical schools include genetics in their curricula as one of the basic sciences that medical graduates must be able to apply. In the application of basic sciences, medical graduates must be able to justify the selection of appropriate investigations for common clinical cases and explain the fundamental principles underlying these techniques. They must also make accurate observations of clinical phenomena and appropriate critical analysis of clinical data.

The Government notes with interest the Committee's further recommendations on training. This includes the introduction of genomic medicine as a core competency in the certificate of completion of training of junior doctors training in the medical and pathological specialties, inclusion as a clinical competency within Continued Professional Development (CPD), and the call for the Nursing and Midwifery Council to set detailed standards across the curriculum on genetics and genomics. It recognises the key importance of education and training in this area. We will, therefore, ask DH, in partnership with the HGSG, to explore with the Postgraduate Deans of Medicine and Medical Education for England, relevant Royal Colleges, the General Medical Council, Postgraduate Medical Education and Training Board, the Nursing and Midwifery Council and other relevant stakeholders, how such training should be organised and recognised.

20. We recommend that the Department of Health should review provision of genetic counselling with regard to both single-gene disorders, single-gene subtypes of common diseases and common diseases (Paragraph 8.48).

On the basis of the findings of the review, we recommend further that the Department should take steps to ensure that adequate provision for genetic counselling is made available within the Regional Genetic Centres and also outside the Centres. The review should take account of the increasing need to support non-specialist physicians in giving accurate and informed advice to patients, and their families, following diagnosis of a single-gene subtype of a common disease (Paragraph 8.49).

The review should also consider the content and scope of training courses for genetic counsellors to ensure that they are able to provide advice on single gene subtypes of common diseases as well as single-gene disorders; and give consideration to statutory professional regulation of genetic counsellors (Paragraph 8.50).

The Government, through commitments made in the White Paper, has already funded, or is in the process of funding, more than 50 NHS genetic counsellor trainee posts. These counsellors work throughout the NHS. The programme is a 'work in progress' and a review at this time would be inappropriate. However, future provision of NHS genetic counsellors and their training will be reviewed before completion of the current scheme in 2011-12.

Genetic counsellors in the NHS already work both independently and with consultant geneticists to provide a high quality and appropriate genetic counselling service to individuals and their families regarding the implications of a genetic diagnosis and genetic testing for both single gene disorders and the single gene subset of common disorders

The Association of Genetic Nurses and Counsellors (AGNC) has developed a process for standardised education and training of genetic counsellors. Practitioners need a background in either nursing or midwifery or need to have completed a master's degree in genetic counselling, with a substantial clinical component, to be eligible to register.

The AGNC has made an application for genetic counsellors to be statutorily regulated by the Health Professions Council (HPC). Once the HPC has considered the application, it will make recommendations to the Secretary of State for Health as to the group's state of readiness for regulation. It will then be for the four UK health departments to make a final decision as to whether it is necessary to regulate genetic counsellors. The regulation of new professions is a devolved matter. However, all four countries are currently committed to UK-wide regulation

21. We recommend that the Department of Health reviews the NGEDC role, to establish whether it has the appropriate structure and mechanisms in place to provide national leadership in training the general medical and nursing workforce in the practice of genomic medicine and the use of genetic testing in the context of common diseases. The aims of the review should be to establish a national programme of training in genomic medicine for the non-genetic medical and nursing specialties, either under the auspices of the NGEDC or another body (Paragraph 8.51).

We recommend that, as part of the current review of the healthcare scientific workforce, the Department of Health should consider how members of the current healthcare science workforce can be trained to enable them to use the new genomic technologies and how to develop bioinformatics skills in particular (Paragraph 8.52).

The Government will give detailed consideration to the Select Committee's recommendation to review the role of the NGEDC.

Currently, DH monitors the work of the NGEDC through the NGEDC Steering Group. In 2009 Professor Charles Easmon was appointed Chair of the Steering Group with a remit from DH to review the functions and working of the Steering Group and its role in developing the strategic vision for the NGEDC. Once this work is completed, early in 2010, further discussions will be held which will take into account the Select Committee's recommendation.

Modernising Scientific Careers, led by Professor Sue Hill, the Chief Scientific Officer, is a key work programme within DH designed to ensure flexibility, sustainability and modern career pathways for healthcare scientists, fit to address the needs of the future NHS. A pilot genetics training programme began in October 2009 with a modernised genetics curricula, which combines both clinical molecular and clinical cytogenetics disciplines, including the development of bioinformatics input. Thirty-two trainees will participate in the pilot on two programmes: Healthcare Scientist Practitioner Training and the Scientist Training Programme. As with other parts of the initiative, this programme will support healthcare scientists in meeting the challenges of new technologies and safeguarding and enhancing the sustainability of this workforce by changing existing training and career arrangements to meet current and future needs.

Future investment in new training places will directly respond to the demand for more genetic tests. This has already increased significantly in the last 10 years, as scientific discoveries have created new opportunities to diagnose and predict disease.

22. We support the Department of Health's commitment to establish a Centre of Excellence for national planning and commissioning of workforce supply and demand. We recommend that the Centre is the appropriate body to provide advice to the NHS on what measures can be taken to address the pressing need to recruit bioinformatics expertise into the service (Paragraph 8.53).

We recommend that the Centre should be asked also to evaluate the workforce planning implications of an expansion of genetic and genomic test services into mainstream specialties (Paragraph 8.54).

A procurement process is underway to establish the Centre for Workforce Intelligence (previously known as the Centre of Excellence) and is scheduled for completion in late 2009, with the Centre becoming operational in 2010. The work programme and priorities for the Centre for Workforce Intelligence will be agreed with DH which will consult key stakeholders through the HGSG.

ACRONYMS AND GLOSSARY

| | |
|----------|---|
| ABI | Association of British Insurers |
| ABPI | Association of the British Pharmaceutical Industry |
| AGNC | Association of Genetic Nurses and Counsellors |
| BIGTR2 | <i>Review and Refresh of Bioscience 2015</i> report |
| BIS | Department for Business, Innovation and Skills |
| CfH | Connecting for Health |
| CPD | Continuing Professional Development |
| DCTs | Direct-to-consumer tests |
| DIUS | Department for Innovation, Universities and Skills |
| DMuDB | Diagnostic Mutation Database |
| DNA | Deoxyribonucleic acid |
| DH | Department of Health |
| DPA | Data Protection Act |
| EASIH | Eastern Sequencing and Informatics Hub |
| EBI | European Bioinformatics Institute |
| EGN | ESRC Genomics Network |
| ELIXIR | European Life Science Infrastructure for Biological Information |
| EMBL-EBI | European Molecular Biology Laboratory – European Bioinformatics Institute |
| ERC | Expert Resource Centre |
| ESRC | Economic and Social Research Council |
| GAIC | Genomics and Insurance Committee |
| GenCAG | Genetics Commissioning Advisory Group |
| GMC | General Medical Council |
| GP | General Practitioner |
| HGC | Human Genetics Commission |
| HGP | Human Genome Project |
| HGS | Human Genome Strategy |
| HGSG | Human Genomic Strategy Group |
| HPC | Health Professions Council |
| HTA | Health Technology Assessment |
| ICO | Information Commissioner's Office |

| | |
|-------|---|
| IP | Intellectual Property |
| IVD | <i>In vitro</i> Diagnostic |
| KTNs | Knowledge Transfer Networks |
| MDD | Medical Devices Directive |
| MHRA | Medicines Healthcare Products Regulatory Agency |
| MISG | Ministerial Industry Strategy Group |
| MMTSG | Ministerial Medical Technology Strategy Group |
| MoJ | Ministry of Justice |
| MRC | Medical Research Council |
| NERC | Natural Environment Research Council |
| NGEDC | National Genetics Education and Development Centre |
| NGRL | National Genetics Reference Laboratory |
| NHS | National Health Service |
| NMC | National Midwifery Council |
| NICE | National Institute for Health and Clinical Excellence |
| NIHR | National Institute of Health Research |
| NMC | National Midwifery Council |
| NPfIT | National Programme for IT |
| OSCHR | Office for Strategic Co-ordination of Health Research |
| PCT | Primary Care Trust |
| PMETB | Postgraduate Medical Education and Training Board |
| PPRS | Pharmaceutical Price Regulation Scheme |
| RCUK | Research Councils UK |
| RAE | Research Assessment Exercise |
| REF | Research Excellence Framework |
| RNA | Ribonucleic acid |
| SHA | Strategic Health Authority |
| SNP | Single Nucleotide Polymorphism |
| TSB | Technology Strategy Board |
| UKGTN | UK Genetic Testing Network |
| WCC | World Class Commissioning |

GLOSSARY

| | |
|------------------------|---|
| Bioinformatics | The application of computers and computational expertise to analyse, visualise, catalogue and interpret large biological datasets in the context of genome sequences of humans and other species |
| Biomedical information | The application of bioinformatics and computational expertise in support of the practice of medicine and the delivery of healthcare |
| Clinical trials | Research study conducted with patients, usually to evaluate a new treatment or drug |
| Clinical utility | The risks and benefits resulting from using a test |
| Clinical validity | The accuracy with which a test identifies or predicts a patient's clinical status |
| Cytogenetics | The study of the relationships between the structure and number of chromosomes and variation in genotype and phenotype |
| Diagnostic tests | A term used to describe particular tests that are able to identify a recognised condition |
| DNA | Deoxyribonucleic acid; the chemical that comprises the genetic material of all cellular organisms |
| Gene | The basic unit of heredity found in chromosomes. A length of DNA that carrier the genetic information necessary for the production of a protein |
| Genetic counselling | Providing an assessment of heritable risk factors and information to patients and their relatives concerning the consequences of a disorder, the chance of developing or transmitting it, how to cope with it, and ways in which it can be prevented, treated and managed |
| Genetic test | An analysis performed on human DNA, RNA, genes and/or chromosomes to detect heritable or acquired genotypes |
| Genome | The unique genetic code or hereditary material of an organism, carried by a set of chromosomes in the nucleus of each cell |
| Genomic medicine | The use of genetic information and genomic tools to determine disease risk and predisposition, diagnosis, prognosis, and the selection and prioritisation of therapeutic options |
| In vitro | (Latin: within the glass). This term refers to experiments performed in an artificial environment like a test tube or culture media |

| | |
|---------------------|--|
| Mutation | A change to the nucleotide sequence of the genetic material of an organism |
| Nucleotide | One of the building blocks of DNA or RNA. There are four nucleotides in DNA: Adenine (A), cytosine (C), guanine (G), and thymine (T). These are the 'letters' or 'bases' of the genetic code |
| Ribonucleic acid | A chemical that is copied from the DNA on an individual's chromosomes, that carries the genetic information required to produce cellular proteins |
| Stratified medicine | The targeting of healthcare interventions, particularly drug treatments, to well-defined subgroups of patients |



information & publishing solutions

Published by TSO (The Stationery Office) and available from:

Online

www.tsoshop.co.uk

Mail, Telephone, Fax & E-mail

TSO

PO Box 29, Norwich, NR3 1GN

Telephone orders/General enquiries: 0870 600 5522

Order through the Parliamentary Hotline Lo-Call 0845 7 023474

Fax orders: 0870 600 5533

E-mail: customer.services@tso.co.uk

Textphone: 0870 240 3701

The Parliamentary Bookshop

12 Bridge Street, Parliament Square

London SW1A 2JX

Telephone orders/General enquiries: 020 7219 3890

Fax orders: 020 7219 3866

Email: bookshop@parliament.uk

Internet: <http://www.bookshop.parliament.uk>

TSO@Blackwell and other Accredited Agents

Customers can also order publications from:

TSO Ireland

16 Arthur Street, Belfast BT1 4GD

Tel 028 9023 8451 Fax 028 9023 5401

ISBN 978-0-10-177572-4



9 780101 775724