House of Lords Science and Technology Committee Report ‘Genomic Medicine’

Request for update on

1. actions taken in response to recommendations
2. original government response to the report

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)

Original Government response:
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.

**Government update (November 2012):**
Since the Government’s original response, the potential offered by the application of genomic technologies for improving healthcare and supporting economic growth has received growing attention. Several workstreams in this area are currently underway.

**OSCHR and Biomedical Catalysts**
Innovation and translation in health care and life sciences are at the heart of the investment strategies of the OSCHR funding partners and the UK government’s plan for growth. The MRC and NIHR, in coordination with OSCHR partners and others, continues to invest heavily in translational research, including through the development of specialist funding initiatives. In December 2011, the Prime Minister launched the UK Life Sciences Strategy, which detailed numerous existing and new initiatives and activities in the translational research arena.

A key announcement was the formation of the TSB/MRC Biomedical Catalyst, a £180m programme, which supports academic and industry scientists to move their research more quickly from discovery to commercialisation. It will particularly focus on taking initial research from Universities through to small and small and medium-sized businesses (SMEs), The first awards of £10m were announced in August with another
£39.1m of funding awarded in October. This brings a total of £49m of funding committed to 64 projects.

**Human Genomics Strategy Group**

The Human Genomics Strategy Group (HGSG) was set up in May 2010 under the chairmanship of Professor Sir John Bell. Its discussions were informed by three working groups each covering a distinct workstream – service development (chaired by Dr Ian Barnes), innovation (chaired by Professor Sir John Burn) and education, engagement and training (chaired by Professor Charles Easmon). An additional focus group (chaired by Professor Dame Janet Thornton) was set up to make recommendations on bioinformatics capacity and capability. The working group discussions, held throughout 2010 and 2011, informed the HGSG’s report which was published in January 2012.

The report is available on the DH website:

The report outlines a vision for genomics in healthcare and makes six overarching recommendations in areas reflecting the working groups’ remits. The report was welcomed by the then Secretary of State for Health and the Minister for Universities and Science who have asked for a framework to implement its recommendations.

Responsibility for implementation will need to reflect the new healthcare governance structure, in particular the establishment of the NHS Commissioning Board and Health Education England.

**BIS/HSBD**

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)*

**Original Government response:**

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.

**Government update (November 2012):**
The Government continues to be committed to embedding the principles of proportionate, risk-based regulation across all regulated sectors.

The Government actively lobbied the Commission and other Member States in recent years to ensure less burdensome legislation for researchers. The new proposal for a Clinical Trials Regulation, was published in July 2012, has many of the elements that the Government lobbied for. The Foreign Secretary referred to the efforts of the Medicines and Healthcare products Regulatory Agency (MHRA) as an excellent example of early influencing.

The Regulation supports a more risk proportionate approach to regulation of clinical trials and allows for coordinated approvals of multinational trials and co-sponsorship of research, with improved approaches to safety reporting. The Government believes that the Commission’s proposal has the potential to make the EU an attractive place for the conduct of clinical trials again. The draft Regulation has been welcomed as addressing many of the issues arising from the current legislation although there are areas where further changes may be needed. The MHRA is actively participating in the ongoing negotiation of the draft Regulation in the Council Working Party and liaising with other Members States, the Commission and the European Parliament and UK stakeholders to ensure that the clinical trials legislation is fit for purposes, risk-based and not unnecessarily burdensome on researchers. MHRA and MRC are working with other funding partners, research organisations and industry representatives to help inform the UK position on the discussions of these Regulations in Europe.

**BIS to clarify/add/delete:**
[Action 26 of Strategy for UK Life Sciences – Streamlining regulation (p27): relevant?]  
A ministerial Star Chamber meeting (DH/BRE) took place on 25 October 2012 [DN – we need to follow up on action from this.]

**BIS (MRC)/Sandor Beukers (MHRA)**

3. We recommend that the proposed White Paper on genomic medicine 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)

**Original Government response:**
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.

**Government update (November 2012):**

The Government’s original response referred to early work on the establishment of Therapeutic Capability Clusters, an 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.

The Therapeutic Capability Clusters pilot initiative was initiated via a call for applications issued in February 2010. The Office for Strategic Coordination of Health Research managed the pilot initiative. In April 2011, as announced in the Government’s *Plan for Growth*, the NIHR Office for Clinical Research Infrastructure (NOCRRI) became responsible for transforming the pilot initiative into a long-term programme – the NIHR Translational Research Partnerships (TRPs). In October 2011, the NIHR established two TRPs in joint and related inflammatory disease and in inflammatory respiratory disease. The TRPs bring together world-class investigators in the UK’s leading academic and NHS centres to support collaboration with the life sciences industry in early and exploratory development of new drugs and other interventions.
In addition, the Biomedical Catalyst has been established to support the transition between academic-led to business-led research and thereby facilitating translational research.

The UK Strategy for Life Sciences, published in December 2011, included a number of initiatives in support of collaboration. The MRC Stratified Medicine programme (£60m) is aimed at developing UK-wide disease specific research consortia to stratify the disease and gain a deeper understanding of the underpinning mechanisms of stratification.

Another initiative is Research Councils UK’s "Principles for Funding Multi-Institutional Collaboration in Innovation and Research" which was published in early 2012. [http://www.rcuk.ac.uk/research/Pages/principles.aspx.]

The appointment of Chris Brinsmead and Professor Sir John Bell as Life Science Champions was made in December 2011. [DN BIS: – be nice to include a sentence about their impact – please supply line]

In order to support science skills in industry, Cogent has undertaken extensive research on industrial placements in the UK and published an evidence report in June. On the basis of this evidence, Cogent is looking to develop a not-for-profit service for industrial placements and mentoring called Developing Tomorrow’s Scientists Today.

The Research Excellence Framework (REF) is being implemented by the Higher Education Funding Council for England (HEFCE) working with devolved HE funding bodies. In March 2011, following detailed consultations from 2007-10, HEFCE confirmed the planned approach to REF 2014, including the assessment of impact from excellent research. An impact pilot exercise conducted in 2010 helped to develop a robust impact assessment methodology, which would be broadly applicable across all disciplines. REF2014 should encourage collaboration between institutions by recognising respective contributions to collaborative research, and should further encourage academics to work with and spend time in industry. “Impact” assessment will account for 20% of REF 2014. The outcomes from the REF will be published in early 2014, and will inform research allocations from 2014-15 onwards.

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)

What have NICE done to further develop a single evaluation pathway for medical and diagnostic technologies?

What consideration is given during the evaluation of such tests to cost savings that traditionally fall outside the cost per ‘QALY’ assessment? For example, how are the additional costs and benefits to patients that occur throughout their lifetime and relate to other treatments and the benefits to family members taken into consideration, where appropriate?

Original Government response:
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.

**Government update (November 2012):**
Since 2009, projects concerning the clinical utility and validity of genetic and genomic tests have been approved for funding by both the NIHR Health Technology Assessment programme and the Efficacy and Mechanism Evaluation programme.

**Single evaluation pathway**
NICE’s Medical Technologies Advisory Committee (MTAC) has been running since 2010. Its purpose is to provide a single entry point to NICE for those developing and using medical and diagnostic technologies. The committee selects medical devices and diagnostics for which NICE should produce guidance and then routes the selected technology to the appropriate NICE guidance producing programme. NICE established two new programmes in 2009 and 2010 - the Medical Technologies Evaluation Programme (MTEP) and the Diagnostics Assessment Programme (DAP) - to carry out these evaluations, and developed bespoke methods and processes for the programmes that focus on the features of innovative devices and diagnostics. The programmes were planned and developed with input from the main industry bodies. Their overall aim is:

- to promote faster uptake of new medical technologies in the NHS;
- to encourage collaborative research, in both industry and the NHS, to generate evidence on the clinical utility and/or healthcare system benefits of selected technologies.
The programmes are fully operational and guidance has been published across the two programmes on a wide range of products, including a device for treating chronic wounds, a contrast agent for imaging the liver, an ultrasound device for scanning burn thickness, a device for self-care in patients with cancer enabling them to manage their own malignant ascites, and genetic tests for familial hypercholesterolaemia.

In addition to developing guidance, NICE uses the programmes to engage with companies so that they can consider and refine the clinical utility of their products for the NHS in advance of evaluation by NICE. NICE also has a research facilitation service run by its External Assessment Centres whereby further evidence is developed on products that have been evaluated by NICE as promising, but where gaps in the evidence remain.

In the 2011 Strategy for Life Sciences NICE was asked by the Department of Business, Innovation and Skills to extend the Scientific Advice service to medtech companies and investors. In response to this NICE has developed a workshop based on developing approaches to value in medtech products, and the first series of 5 workshops has recently been held, receiving extremely positive feedback. NICE will provide more of these in response to demand.

NICE regularly meets both individual medtech companies and industry bodies as part of their engagement over the programmes. Feedback from companies that have had their products evaluated by NICE, and from industry bodies on the work of the programmes is generally extremely positive.

**Evaluation**

NICE’s approach to evaluating diagnostics involves estimating the outcomes that the patient will experience as a result of using the diagnostic technology, and estimating the costs to the healthcare system. Added complexity arises because more extensive modelling is usually required to include the initial testing, follow-up testing, treatment, monitoring and capturing down-stream effects. The outcomes and costs typically include those arising from treatments following the use of the technology and cover the entire relevant portion of the care pathway. Therefore, in principle, NICE’s evaluation approach will incorporate important downstream effects arising from a genetic test diagnosis. Data allowing valuation of patient preferences in terms of potential reassurance and non-health benefits provided by knowledge of test results are not always available and in these cases, the decisions are made by deliberation the committee whose members include Specialist Committee Members and lay members with knowledge of the condition and the potential wider impact of the diagnostic.

The economic modelling takes account of all NHS and personal social services costs. Costs incurred by patients and carers are not included in the economic modelling but may be considered, alongside patient preference issues by the medical technologies advisory committee in its decision making.

*UK Genetic Testing Network*
Complementing NICE activities, the UK Genetic Testing Network undertakes the evaluation of tests that do not meet the selection criteria set out by NICE due to low volume of these tests and subsequent limited evidence base. This process is undertaken through the UKGTN Genetic Test Evaluation Working Group on an annual cycle to fit with the contractual commissioning arrangements. The UKGTN Clinical and Scientific Advisory Group consider the recommendations at its meeting in September and if endorsed the new tests are incorporated into the UKGTN Directory of Genetic Testing in the following April. Since July 2009, 171 new tests have been recommended for adoption in the NHS. Commissioners are advised of the costs of the new tests, the changes in the diagnostic and treatment care pathways that could lead to savings (by stopping investigations and interventions) the benefits of the new tests for patients and their families and the impact on management/treatment where appropriate. This information is presented annually in October to the specialised services commissioners for potential funding from April the following year and to NICE to review the implications on mainstream services such as cardiology and cancer services. Information is published on the UKGTN web site (Gene Dossiers, Testing Criteria and the laboratories providing the tests) so it is open and transparent for clinicians and the public. The NHS Commissioning Board (NHSCB) recognises this process and has built the work of the UKGTN into the service specification for medical genetics.

Jonathan Bickley/Carl Glenister/Jacquie Westwood/Jane Deller

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)

What progress has been made regarding the potential reclassification of genetic and genomic tests as medium risk in the revision on the EU In Vitro Diagnostic Medical Devices Directive?

Original Government response:
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.
Government update (November 2012):
On 26 September, the European Commission adopted its proposal for a new regulation on in vitro diagnostic medical devices (IVDs). The Commission has proposed to move from a classification system that uses specific lists of IVDs to a risk-based classification based on more general rules in line with global guidance.

They have also proposed to amend the definition of an IVD to clearly include genetic tests and that they are classified as medium risk (Class C) devices. This means that the manufacturer’s quality assurance system and design dossier will be scrutinised by a notified body before they can place a genetic test on the market. The UK will be supporting this change.

Graeme Tunbridge/Rebecca Brown

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)

What action have Government taken to ensure that the new Pharmaceutical Price Regulation Scheme (PPRS) will reflect the true value of stratified medicines to a patient’s health and wellbeing throughout their lifetime?

What action has been taken to introduce the option of flexible pricing structures that allow pricings for stratified medicines to be adjusted according to the added value that they bring to a patient?

For example, when additional stratification of a patient group results in a more cost effective targeting of the use of medicines?

Original Government response:
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 (Erbitux®).

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.

**Government update (November 2012):**
The 2009 Pharmaceutical Price Regulation Scheme (PPRS) included a new flexible pricing mechanism, allowing companies to propose an initial price for a medicine that reflects value at launch, while retaining the possibility to apply
for an increase or decrease to this original list price either as further evidence or as new indications for the medicine emerge and change the effective value that the medicines offers to NHS patients. To date, there have been no applications under the flexible pricing mechanism, though it remains open for the duration of the 2009 PPRS.

The Government is working towards a new system of pricing for innovative medicines, including value-based pricing, from January 2014, following the end of the current PPRS. Under value-based pricing, the price the NHS pays for a medicine will be linked to the value that a new medicine delivers, i.e. the benefits doctors and patients will see from a drug.

In developing the value-based pricing system, the aim is to create a system that has the capability to include the broadest possible range of new medicines, thus minimising the need for parallel mechanisms.

Katy Peters and colleagues

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). 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)

Original Government response:
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.

**Government update (November 2012):**
Since our original response, the Government has supported a number of initiatives to promote the development of stratified medicines.

For example, as part of its work in establishing the Stratified Medicine Innovation Platform (SMIP), the TSB and partners MRC, DH England,
Scottish Executive Health Directorate, Cancer Research UK, Arthritis Research UK, and the National Institute for Health and Clinical Excellence (NICE) developed and published the roadmap for Stratified medicine in the UK.


The roadmap and vision for the UK was built with input from over 100 individuals from key stakeholder groups and identified a number of priorities for the partners in the SMIP partnership to work together to deliver on. The thematic priorities include:

- Incentivising adoption
- Increasing awareness
- Patient recruitment – consents and ethics
- Clinical trials
- Data – collection, management and use
- Regulation and standards
- Intellectual property
- Bio-banks and biomarkers
- Increasing the impact of R&D investment

The SMIP partnerships have been collectively exploring how they deliver interventions within these various thematic areas.

In addition to the TSB-led initiative, the MRC is using a multi-partner consortium approach to address key priorities in stratified medicine – to create understanding in complex areas of importance to industry and academia, in areas jointly agreed, and with jointly-developed research programmes. In 2010, the MRC and ABPI agreed an initial strategic focus on inflammation and immunology, and the MRC invested £9.5m to establish two disease-specific consortia in Chronic Obstructive Pulmonary Disease (COPD) and Rheumatoid Arthritis. To date, 15 companies are involved in the two consortia, from five at the outset of the award. Subsequently, a further £6m was awarded to assemble an industry/academic consortium on disease stratification in Type 2 diabetes. The MRC is now implementing a £60m, four-year MRC investment in stratified medicine research that was announced as part of the UK Life Sciences Strategy in December 2011.

Our previous response referred to the MHRA/ABPI Forum on personalised medicine held in October 2009. A report from the Forum has since been published on the MHRA website

http://www.mhra.gov.uk/Howweregulate/Medicines/MISGNewTechnologiesAdvisoryPanel/Forums/CON065590

Also, in 2011, a follow up conference was organised jointly by the Bioindustry Association (BIA) and the MHRA titled “Personalised medicine – the evolving regulatory landscape” to explore the current regulatory strategies and initiatives in enhancing personalised medicine in UK and in Europe. The meeting was chaired by Dr Ian Hudson and by Dr Alan Morrison and was
attended by several leading figures including Prof Gordon Duff, Chair of CHM, and Professor Munir Pirmohamed and Dr Eric Abadie, the then Chair of CHMP.

Key aspects discussed at this conference included the need for a concerted effort towards application and utilisation of stratified medicine, the opportunities afforded by European regulatory schemes including the biomarker qualification process and the opportunities for developments of pharmacogenetic tests. A discussion on use of parallel or unified strategies for concomitant development of the treatment and the test (companion diagnostic) was also an important part of this conference. The discussion helped inform the regulatory policy and approaches towards the current proposal for the In Vitro Diagnostic Devices Regulation.

The UK (including MHRA) have contributed to the discussions at European level at public meetings and conferences on personalised medicine helping to shape the overall progression of stratified medicine at national and European regulatory levels.

**BIS/Zahid Latif/Ian Hudson**

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)**

**Original Government response:**
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.
Government update (November 2012):
In October 2010, the Technology Strategy Board opened three competitions for funding (subsequently awarding £9.5m of grant) to support the development of technologies and business model in the area of stratified medicine. Also in October 2010, the TSB Governing Board approved the establishment of the Stratified Medicine Innovation Platform to commit £50m over 5 years to support the development of business led projects to pursue stratified medicine approaches in the UK.

http://www.innovateuk.org/_assets/ipbrochure_stratifiedmed_final.pdf

The Stratified Medicine Innovation Platform builds on the UK’s strength within the global healthcare industries and puts it at the centre of the next generation of medicine. The vision is for the UK to be the best place to develop, and have adopted, stratified medicine. The Platform aims to help accelerate the rate of development and uptake of stratified medicines in the UK, for the benefit of patients, healthcare providers and business. It is a partnership of seven organisations that have agreed to work together and combine resources to achieve this:

• Technology Strategy Board
• Medical Research Council
• Department of Health England
• Scottish Executive Health Directorate
• Cancer Research UK
• Arthritis Research UK
• National Institute for Health and Clinical Excellence (NICE)

The seven partner organisations will together invest around £200m over five years in the area of stratified medicine. The investment will go into innovative technological R&D in areas such as improved tumour profiling in cancer, accelerating the validation and adoption of biomarkers, and the uptake of medicines and companion diagnostics in the NHS.

An example of how the programme partners have worked together includes the TSB competition ‘Tumour profiling and data capture to improve cancer care’ which was launched in October 2010 to help companies develop test systems that can examine many different genetic mutations at the same time. The tests are aimed at a potential market being created by Cancer Research UK’s stratified medicines programme to test around 9,000 patients newly diagnosed with one of six tumour types (breast, bowel, lung, prostate, ovary and melanoma) for a number of common gene mutations, and to use the tests to guide treatment and monitor outcomes. They aim to show that standardised genetic testing of tumours on a large scale is feasible, and to collect research data to improve treatment targeting in the future. The Cancer Research UK programme is investing around £5.5m programme to pilot routine tumour profiling.

This is an extremely important step: 280,000 new cases of cancer are diagnosed every year in the UK, and there are increasing numbers of genetic-
targeted treatments in use and in development. A second strand of the competition aims to help companies develop data-handling solutions that will allow Cancer Research UK to provide data from these samples to researchers and doctors to help develop new therapies for the future. In total £5.8m has been invested in six projects, four developing assay technology, two dealing with data-handling solutions.

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 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, 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)

What assessment has been made of the disparity in access to genetic and genomic tests across the UK? (For example, tests for the diagnosis of single-gene disorders, genetic subtypes of common diseases or pharmacogenetic tests). What actions have been taken to ensure that the “postcode lottery” for the provision of NHS genetics and genomic tests across the UK has been remedied?

Original Government response:
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 servicedelivery 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.

**Government update (November 2012):**

*Measuring genetic testing rates*

The UKGTN has undertaken reviews of UK genetic testing rates since 2007. The results are presented annually to the UKGTN Clinical and Scientific Advisory Group. The work is supported by the London Health Observatory to map the results by geographical region. To date the rates have been mapped to Strategic Health Authorities and Primary Care Trust geographical regions. In the future, the rates of genetic testing will be mapped to the new geographical boundaries being established by the NHS Commissioning Board (NHSCB), including Specialised Commissioning Local Area Teams, NHS CB Regional Offices, Clinical Commissioning Groups and the equivalent boundaries of the devolved countries. The quality of the data is improving over time as demonstrated by the trend analysis. NHSCB has recognised the importance of this work and has included a mandatory requirement in the Medical Genetics service specification to submit data for this UKGTN work. It is expected that the development work undertaken will have sufficiently progressed by next year so that it can be published. This work is being fed into the E-Atlas so that comparative tables can be established to inform commissioners and provider laboratories of any variations.

The NHS Atlas of Variation series is published by NHS Right Care and is intended to support local decision making to increase the value which a population receives from the resources spent on their healthcare. It supports the search for unexplained variations, the identification and attention to
unwarranted variation, helping clinicians to understand what is going on in their area and where to focus attention to improve the care they provide. The first NHS Atlas of Variation was published in November 2010, with topics selected by clinicians as being important to their speciality. In December 2011 a second and expanded version of the Atlas, consisting of 71 maps, was published and this included discussion on extending the atlas to include pathology. The Atlas will enable commissioners to review variations and take action where required depending on the local population needs.

Promoting equity in commissioning
Separate to the review of genetic testing rates, and in order for activity across laboratories to be compared for commissioning purposes, the UKGTN in collaboration with professional groups has developed a common laboratory currency. The system, termed Genetic Units (GenUs), allows molecular and cytogenetic laboratory activity to be compared across providers in the UK. The GenUs activity measures have been adopted by the specialised commissioners and will be used in future NHS Contracts with Providers.

UKGTN has been working with the NHSCB Medical Genetics Clinical Reference Group to develop consistent commissioning policies for implementation in April 2013. This has included a common service specification; a quality dashboard (information being collected from quarter 3 in 2012/13); QIPP schemes; quality, innovation, prevention and productivity and CQUIN schemes and quality and innovation initiatives. These documents are currently being quality assured through the NHSCB Commissioning Assurance Group. The implementation of these initiatives is expected to regularise commissioning arrangements over a two year period so that the commissioning arrangements are uniform across England thus reducing variations in practice.

Within Scotland, single gene and genetic sub group testing services are nationally designated and funded through the Scottish Genetic Laboratory Consortium which is commissioned and managed on behalf of NHS Scotland by National Services Division (NSD). This is the mechanism used to ensure equitable access for residents of Scotland. From 1 April 2013, pharmacogenetic testing using molecular pathology techniques will also be nationally designated, and funded centrally through NSD. A decision-making framework to evaluate pharmacogenetic tests is being developed and it is expected that this will be aligned with the work underway by the National Institute for Clinical Excellence.

Jacquie Westwood/Deirdre Evans

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)
**Original Government response:**
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.

**Government update (November 2012):**

The NICE Diagnostic Assessment Programme is taking forward reviewing companion diagnostics for pharmacogenetics. UKGTN is working with NICE to share information about test evaluations to enable the diagnostic assessment board to consider whether NICE guidance for particular conditions would be warranted. UKGTN is holding workshops for particular conditions in order to develop further Testing Criteria where practice is informed by new developments such as for Marfans and neurological conditions. For cancer developments, UKGTN has liaised with the Cancer Genetic Group so that any new tests being reviewed are also considered by the expert cancer geneticists to inform any recommendations.

Jacquie Westwood

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)
What progress has been made on bringing together molecular pathology and genetics laboratory services; particularly to take forward the HGSG’s recommendation to develop a network of Genomic Technology Centres, Biomedical Diagnostic Hubs and Regional Genetics Centres?

Original Government response:
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.

Government update (November 2012):
To be completed

Christine McCartney:
“Pathology Modernisation, which is the rationalisation, consolidation and transformation of these core services, is happening across the country at an increasing intensity and the HPA regional Public Health laboratories are all involved in this. The HPA and PHE has made a commitment to having a network of public health laboratories and the HPA is pursuing this strategy in conjunction with its NHS “partners” in the major conurbations in which it is located. Our overall aim is to ensure that we emerge from this modernisation phase of the NHS with a resilient network of public health laboratories appropriately located in major population centres to deliver the outputs needed by PHE.”

As part of this exercise we are actively working to establish synergistic relationships between molecular microbiology/pathology and genetics laboratories in the key regional locations.

MRC is scoping the delivery of molecular pathology for medical research and will report to OSCHR on potential approaches to this in 2013.

Ian Barnes/HPA/MRC

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)

Original Government response:
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.

Government update (November 2012):
European partnership in the EBI provides resources and specialist expertise to help deliver against the growing need to collect, store and curate huge amounts of data. MRC invests around £12m p.a. for the UK’s subscription to EMBL, which in turn provides long-term, sustainable core funding to EBI (ca £20m p.a. across all partners). Renewed funding from the 21 EU Member States for EMBL (2012-2016) was agreed at the end of 2011.

Building on this investment the UK has invested in the ELIXIR data centre and technical Hub. In 2011, the UK Government invested £75m capital funding in the ELIXIR Technical Hub project and Data Centre Capacity project at the EBI. The European Molecular Biology Laboratory’s European Bioinformatics Institute (EMBL-EBI) in the UK is coordinating ELIXIR. The purpose of ELIXIR is to construct and operate a sustainable infrastructure for biological information in Europe to support life science research and its translation to medicine and the environment, the bio-industries and society. ELIXIR will construct, operate and enhance a distributed research infrastructure in accordance with the requirements of the scientific community and under the direction of the ELIXIR Board. The ELIXIR Hub will be connected to ELIXIR Nodes to provide infrastructure for data, compute, tools and standards and training. The UK Node for ELIXIR is currently being developed with focus on training specifically to develop informed users and providers of software, data and services.
Funding for ELIXIR Large Facilities Capital Fund Programme was confirmed in the ‘Strategy for UK Life Sciences’ and Willmot Dixon commenced construction of the Technical Hub facility in June 2011. The programme is progressing well, it is within budget and on target for the required BREEAM excellent rating. Completion is due by October 2013 and a topping out ceremony is being planned for February 2013.

BIS

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)

What progress has been made in putting in place appropriate structures to build excellence in bioinformatics across the service?

Original Government response:
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

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.

Government update (2012):
The Human Genomics Strategy Group recommended that the Department of Health in partnership with BIS and other relevant partners should develop proposals to establish a central repository for storing genomic and genetic data, and relevant phenotypic data from patients, with the capacity to provide biomedical informatics services and an open-data platform that SMEs can build on.

We are working with our partners on taking this recommendation forward.

As stated in our original response, the Government recognises the importance of linking medical and genetic information if the potential offered by genomic medicine is to be realised fully. The Government's activities in the area of data sharing are set out under paragraph 16.

Supporting research
From a research perspective, the Government has already invested in the development of data linkage services to support life sciences research – the Life Sciences Strategy announced investment in the Clinical Practice Research Datalink (CPRD) (for more detail on CPRD see below). The linkages service is in early discussions to establish a safe and secure mechanism to incorporate genomic data to then provide suitable research data to support life sciences research.

In addition, it is clear that to remain a global player in research, substantive capital and recurrent investment is required to develop a framework for a national biomedical informatics infrastructure including for example the co-localisation of data, compute power, software, training and expert support. Such a framework is under discussion as part of the RCUK ‘Investing for Growth’ capital investment strategy and would complement existing and planned investments, for example those at the European Bioinformatics Institute, the UK’s contribution to ELIXIR, and the Department of Health’s development of health data sets and related informatics, as outlined in the Life Sciences Strategy (Dec 2011).

More specifically, MRC invested £10m in 2009 in four High Throughput Sequencing Hubs to enhance the UK capabilities in next generation high-throughput sequencing and associated bioinformatics. Skills and capacity building are crucial to this endeavour and MRC has additionally invested ca.
£14m since 2009 in specialist training programmes at Masters, Post Doctoral, and early Independent Investigator levels in the field of bioinformatics, computational biology and medical statistics. Further investments in these areas are also supported through our Institutes/Units and grant funding.

Developing formalised and commissioned education and training for bioinformatics professionals
As a matter of urgency, curricula are being developed to introduce a training programme for bioinformatics as one of the new pre-registration postgraduate scientist educational programmes within Modernising Scientific Careers. This will lead to both an academic and work based blended learning programme at Masters-level, with delivery overseen by the National School of Healthcare Science, and is anticipated to lead to registration with the Health and Care Professions Council (HCPC) as a clinical scientist. It will include a year of rotational training including computer programming and health informatics followed by two years of specialist training in genomics and bioinformatics. A specialist working group has been convened to take this forward and work has begun around workforce planning and commissioning Masters academic programme from the Higher Education sector for a 2013 academic year start.

Embedding training in genomics for the wider healthcare workforce
The National Genetics Education Resource Centre in conjunction with the National School of Healthcare Science based in the West Midlands Medical deanery is both reviewing existing education and training provision for healthcare professionals and undertaking a training needs assessment to ensure that appropriate modules can be embedded. This will include the more specialist workforce in pathology and genetics

Exploring the links with other bodies
There is a wealth of information available that is being identified and assessed for CPD and other purposes. The relationships to a future bioinformatics function and to other emerging structures such as the Academic Health Science Networks are also being explored.

BIS/Jonathan Bickley/ Sue Hill/Pat Saunders

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)

Original Government response:
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.

**Government update (November 2012):**
The IT needs around bioinformatics and genomics have been recognised and we are exploring with our partners how these issues can be addressed.

Mike Curtis

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)

Who will be responsible for the activities previously allocated to the Human Genetics Commission, including:
- public engagement activities on the ethical, legal and social issues associated with further integration of genomic technology into mainstream healthcare provision;
- the provision of independent advice to Government on human genetics and genomics with a particular focus on the social, ethical and legal issues

Original Government response:
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.

**Government update (November 2012):**

Since the original Government response, BIS has committed to the continuation of the Sciencewise Expert Resource Centre until 31 March 2015 and the Programme is in discussion with DH regarding opportunities to ensure public engagement and dialogue on genomic issues.

As a result of the Department of Health’s review of its Advisory Non-Departmental Public Bodies, the Human Genetics Commission was disbanded. A new departmental advisory committee, the Emerging Science and Bioethics Advisory Committee (ESBAC) is now the main UK advisory body on emerging healthcare scientific developments, including genetics and genomics, and their ethical, legal, social and economic implications. It is sponsored by the Chief Medical Officer (CMO) for England and chaired by Professor Sir Alasdair Breckenridge. Its membership includes representation from all UK Health Departments, to whom ESBAC also provides advice. The purpose of ESBAC is to provide expert advice to support DH policy development and priority setting in healthcare science – if tasked, this may include advising DH on genetics and genomics issues. ESBAC will also operate as a networking forum to exchange information about matters
relevant to its remit from Members, nominating organisations and wider stakeholders.

Whilst the scope of the work of ESBAC will have to remain by definition broad, ESBAC will not cover all bioethics issues. It will only address concerns arising from major new scientific and technological developments and their relationship to healthcare. Aspects of bioethics that are currently covered by other organisations, or are long-running ethical debates are therefore out of scope.

The Committee has just met for the second time to finalise its Code of Practice, develop an approach for horizon scanning and progress its workplan.

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 so as to facilitate the conduct of non-personal research using genetic data (Paragraph 8.33)

Original Government response:
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.

**Government update (November 2012):**
The Department of Health is tackling changes in the legal frameworks and being proactive with the population on what to expect from the NHS in respect to the use of patient records to support life sciences research.

There are a number of updates on the above recommendations:

*Safe Havens*
The Health and Social Care Act 2012 creates a strategic safe haven, the Health and Social Care Information Centre (HSCIC), for data to support
research and other key work requiring the processing of health and social care data, including potentially genetic data.

From April 2013, the Health and Social Care Information Centre will take on its new status and functions – collecting, linking securely and publishing a wealth of core data – enabling it to become the national focal point and key resource for health and care information. Provisions in the Health and Social Care Act 2012 strengthen and clarify the role of the Health and Social Care Information Centre so that information can be collected, held securely and made readily available to those who need it in safe, de-identified formats, with crucial safeguards in place to protect the confidential data it holds.

The co-ordination of Information and Communication Technologies (ICT) development in the NHS is in transition following the reforms delivered by the Health and Social Care Act 2012. The Government’s Plan for Growth, Life Sciences Strategy and the Government’s Information Strategy for health and social care in England have developed the concept of a ‘safe haven’ – a place to process patient data in a secure and safe way, and deliver anonymised patient longitudinal records to support purposes such as life sciences research. Any further ‘safe havens’ would need to have a clear legal basis to hold and use confidential patient data for specific purposes.

The Clinical Practice Research Datalink (CPRD), launched in April 2012 is working with HSCIC, to provide access to data from linked NHS patient records to produce anonymised high quality research data. CPRD and HSCIC are currently engaged in discussions on how to link genomic data to NHS patient records in a secure and safe way to support life sciences research. These discussions are at an early stage.

The forerunner of the CPRD, the Research Capability Programme, established a safe and secure Information Governance model of operation, which has wide support from independent bodies, the BMA and Information Commissioners Office.

Additionally, the Government, via the NHS Constitution consultation, has made clear with the population the use of NHS data to support life sciences research and other purposes. The consultation (launched 5 November 2012) makes clear that anonymised data is used to support research but where this is not possible then either the individual’s consent or another legal power will be used to access the records. The right of an individual to object to identifiable data being used to support such activities, and have that objection considered, is also be clearly articulated.

_Caldicott review_

There is a need for greater clarity over the circumstances when it is in our interest for our personal health and care information - including our genetic data - to be shared, and for the legal basis to do so in each case. Following a recommendation from the Future Forum on this issue, earlier this year the then Secretary of State, asked Dame Fiona Caldicott to lead an independent review of the current information governance rules, and of their application, to
ensure that there is an appropriate balance between the protection of patient information and the use and sharing of information to improve patient care. The Review is looking at the linking of genetic information with other health data, and genetics and genomics data issues more broadly, and is likely to make recommendations in this area. The Review Panel is expected to report its recommendations in 2013. Future use and linking of personal genetic data, including 'safe havens', will need to take account of that review's recommendations.

ICO Data Sharing Code of Practice
In May 2011, the Information Commissioner issued a new statutory data sharing code of practice and is currently finalising a new Code of Practice relating to the use of Anonymised data. The data sharing Code has been published under section 52 of the Data Protection Act and provides a framework for organisations to make good quality decisions about data sharing and is the ICO's interpretation of what the DPA requires when sharing personal data. The Code of Practice covers both routine and one-off instances of data sharing and is designed to help businesses and public sector bodies share people's personal information appropriately. It helps organisations collect and share personal data (including sensitive personal data) in a way that is fair, transparent and in line with the expectations of individuals whose information is being shared. Specific benefits of the Code for organisations include a minimised risk of breaking the law and consequent enforcement, increased data sharing when this is necessary and beneficial, and a better understanding of when, or whether, it is acceptable to share information without an individual's knowledge or consent.

The Code covers a variety of activities including:
- a GP sending information about a patient to a local hospital;
- two neighbouring health authorities sharing information about their employees for fraud prevention purposes;
- a school providing information about pupils to a research organisation.

The Department of Health has provided modules to its [DN: the Code’s?] Information Governance Toolkit, an on-line tool for promoting information assurance, for research teams ghosted by academic units and for large organisations [DN: what does this mean?] such as the Health & Social Care Information Centre to ensure that they are supported with clear guidance and also report on their performance in a standardised manner. This is intended to support the concept of a 'research passport' enabling researchers to demonstrate that they meet appropriate information handling requirements.

Proposed EU Data Protection Legislative Framework
The European Commission published new legislative proposals for data protection on 25 January 2012 – the Ministry of Justice is leading on the UK response to this. The proposals contain a draft Regulation (setting out a general EU framework for data protection) and a draft Directive (covering authorities dealing with criminal offences and penalties). If the proposed Regulation comes into force, it will repeal the UK Data Protection Act 1998 to
provide an EU-wide Data Protection legal framework. The Regulation makes a new provision under Article 83 for ‘processing for historical, statistical and scientific research purposes’. This provision provides conditions and safeguards for processing data for research purposes and requires identifiable data to be held separately from other data. The Government wants to see EU data protection that protects the civil liberties of individuals while allowing for proper public protection and economic growth and we welcome a provision for the research use of data. However, there are a number of areas of concern. As part of the negotiating process, MoJ will continue to engage with stakeholders including ICO, DH and the National Archives to ensure that their interests are taken into account in any proposed changes. The Department of Health believes that the key area to life sciences research are the individual’s ‘right to be forgotten’ and just how far this can be implemented practically, and the fine regime that increases the maximum fine from the current UK position of up to £500K to 2% of global turnover, which for life sciences companies could be hundred of millions of Euros.

The Department of Health is supporting the Ministry of Justice to ensure that the interests of the health and social care sectors, including research, are understood and recognised in any proposed changes.

David Knight/Diana Paine/Phil Walker/Joana Morrison

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 above. (Paragraph 8.37)
Who will be responsible for the activities previously allocated to the Human Genetics Commission, including:
- monitoring of the need for specific legislation against genetic discrimination;
- consideration of a long-term solution to the current concordat and moratorium on genetics and insurance that has been extended to 2017?

**Original Government response:**
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.

**Government update (November 2012):**
The Concordat and Moratorium on genetics and insurance continues to work well. During 2011, the Department of Health and the Association of British Insurers (ABI) undertook a planned review of the Concordat to ensure it remained fit for purpose. The Concordat now incorporates provisions of the ABI Code of Practice on genetics and insurance, making it easier for consumers to find all the relevant information in one document. The Concordat also incorporates an improved process for approving applications for use of predictive genetic tests in insurance underwriting should they be made.

The revised Concordat was published in April 2012 and the next review will be in 2014 and at three-yearly intervals thereafter. ABI will monitor the operation of the Concordat and Moratorium and will report on this to DH officials on an annual basis. ABI member firms will have to confirm that they will comply with the Concordat and Moratorium for its full duration. The ABI will publish a list of firms that have confirmed compliance on its website. Each three-yearly review will also consider any other comments or evidence that might impact on the operation of the Concordat and Moratorium. In this way, the Government will continue to monitor the need for specific legislation against genetic discrimination.

The revised Concordat also addresses concerns that the Concordat should offer continued protection to consumers who decide to take a predictive genetic test now (i.e. under the Moratorium) but who may wish to take out insurance at a later date – the so-called ‘test now, buy later’ problem. The agreement has been strengthened: with the ABI giving an undertaking not to seek an end to the agreement outside of the planned review process or before the end of the Moratorium. With the Moratorium extended to 2017 and the next review due in 2014, consumer will have at least three years to prepare for any change.

The Government believes that the Concordat and Moratorium remains a proportionate measure for protecting the interests of both customers and insurers and sees no reason for legislation on the use of genetic test results or family history during the term of the agreement.

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)
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)

Original Government response:
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).

Government update (November 2012):
The Human Genetic Commission’s Common Framework of Principles for direct-to-consumer genetic testing services was published in August 2010. The report is available on the archived Human Genetics Commission website (now archived with the National Archives and the British Library web archive).

The Principles were developed by an HGC-led international expert working group comprising representatives from the genetic testing industry, experts in regulation, clinical and molecular genetics and genetic counselling, representatives from groups that support people with genetic conditions, and the Department of Health.

The Principles are aimed at promoting high standards and consistency in the provision of genetic tests amongst commercial providers at an international level and helping to protect the interests of people seeking genetic tests and their families. The Principles identify areas where individual providers, professional organisations, regulatory bodies, and/or national jurisdictions should have defined measures in place, and the nature of those measures.
They cover all aspects of direct-to-consumer genetic testing services including the marketing of tests, information for consumers, consent, laboratory analysis of biological samples and the levels of support that should accompany the provision of genetic test results.

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)

Original Government response:
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.

Government update (November 2012):
The Department of Health is working with Health Education England to establish a lead Local Education and Training Board (LETB) for genomics education and training. This is building upon the success of the NHS Genetics Education and Development Centre and is linked to the specialist healthcare scientist education and training developments in genetics overseen by the National School of Healthcare Science based in the West Midlands Medical Deanery. A HCSG [DN: should this say HGSG?] work programme has been developed around embedding education and training in genomics in both pre and post registration programmes for all healthcare professionals working with the relevant professional advisory bodies across the different professional groups and undertaking a needs assessment of the training needs.

The MRC Clinical Pharmacology and Pathology Fellowship Programmes aim to provide integrated, in-depth, advanced skills research training in clinical pharmacology and/or pathology as applied to internationally competitive organ, tissue or systems based research, within an outstanding basic,
translational and clinical research environment. In March 2010, two awards were made at a total cost of £3.5m. Together, the two programmes will provide PhD training for 20 clinicians, recruited for starts in 2011 and 2012.

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)

Original Government response:
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.

**Suggested update:**
The Government recognises the important role that genetic counsellors play in the delivery of genetic services. We are aware that genetic counsellors are taking an increasingly independent role in supporting the new genomic technologies including consent, interpreting and explaining complex and unexpected results (within the context of multidisciplinary teams with clinicians, laboratory scientists and bioinformaticians) and carrying out family studies as part of pathogenicity co-segregation studies or to support the impact on the wider family.

Multidisciplinary clinics are increasingly common, with specialised genetic counsellors working across medicine (including cardiac, ophthalmology, and cancer clinics) and genetic counsellors providing training and support for specialist nurses such as cardiac nurses, screening midwives and cancer nurses.

*Not sure what to say about funding for training and registration issues.*

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21. **We recommend that the Department of Health reviews the National Genetics Education and Development Centre’s (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)**

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What progress has been made in determining the workforce planning needs for bioinformatics services in the NHS and putting in place appropriate structures to build the workforce?
What action does the Government intend to take to review existing provision of genomics training and education for each profession that works with patients that require genetic and genomic tests (including medical students and qualified doctors, nursing students and qualified nurses, medical laboratory scientists and genetic counsellors); and to develop an action plan to ensure that workforce developments do not lag behind service developments, as recommended by the HGSG?

**Original Government response:**
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.

**Government update (November 2012):**
In the new educational infrastructure within Health Education England, the lead Local Education Training Board for healthcare science will include a national function covering commissioning of education and training for healthcare scientists. This year all education commissioners have been asked to provide potential numbers – this will then be reflected in education commissions to enrol on the new training programme in bioinformatics in 2013. There continues to be recruitment to the postgraduate scientists training programmes in genetics with the first cohort from the combined cytogenetics and molecular genetics Modernising Scientific Careers programmes exiting this Autumn. All have secured posts.
A new genetics technology undergraduate programme has been established and accredited. A highly specialist formalised 5 year training programme in genetics is being developed in conjunction with the Royal College of Pathology.

See response to recommendation 19 as well.

Pat Saunders/Sue Hill

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).

Original Government response:
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.

Government update (November 2012):
See earlier response.