

**House of Lords Science and Technology
Committee**

Genomic Medicine

**Government Memorandum
April 2008**

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House of Lords Science and Technology Committee

Memorandum of evidence on Genomic medicine, April 2008

Introduction

1. The Government welcomes the House of Lords Science and Technology Committee appointment of a sub-committee, chaired by Lord Patel, to assess the current state of genome technologies and their actual and potential impact on clinical practice.
2. The Committee has expressed an interest in a range of issues related to:
 - the development of a policy framework;
 - the research and development strategy, including that of the research councils;
 - the use and storage of biological data;
 - the translation into clinical practice and potential impact on the NHS
3. This memorandum is in response to the call for evidence, and highlights the cross Government work by the Department of Health (DH), the Department for Innovation, University and Skills (DIUS) and the Department for Business Enterprise & Regulatory Reform (BERR). This memorandum does not cover certain policy areas that are devolved to Scotland, Wales and Northern Ireland, mainly those relating to the funding of research and development in the National Health Service (NHS). However, some initiatives are funded on a UK-wide basis and these are indicated. The memorandum focuses on many of the issues raised by the Committee, by:
 - highlighting the significant initiatives that the government has undertaken to support genomic medicine;
 - giving examples of how some of the initiatives are currently being used and may be used in the future;
 - describing how horizon scanning is carried out and the regulatory considerations.
4. In particular, the memorandum of evidence highlights the work under the 2003 Genetics White Paper *Our inheritance, our future*¹ to develop the capacity and capability of the NHS. It will also focus on *Best Research for Best Health*², which sets out the direction that NHS research and

1

http://www.dh.gov.uk/en/Publicationsandstatistics/Publications/PublicationsPolicyAndGuidance/DH_4006538

2

http://www.dh.gov.uk/en/Researchanddevelopment/Researchanddevelopmentstrategy/DH_4127109

development will take to ensure a world-class environment for conducting and using NHS health research.

5. The Human Genome Project, completed in 2003, resulted in the sequencing and identification of the 25,000 or so genes that make up the human race. Since then a number of other human (and animal) genomes have been completely sequenced thus paving the way to discovering the meaning and impact of individual differences in our genes. This, in turn, will facilitate a greater understanding of how groups of genes interact not only with each other but also with environmental factors to influence resistance to, or development of, disease. These advances in human genetics have the potential to change significantly the healthcare pathways involved in disease prevention, diagnosis and treatment. The application of knowledge of the human genome to medical practice could be defined as genomic medicine.
6. To properly harness this potential for the NHS, will require a comprehensive evidence base. This is being generated by a combination of conventional research and development together with new methodologies that utilise faster analytical techniques. The evidence base will also comprise of evaluations, economic analyses and examinations of the related social, ethical and legal issues. The Government, Research Councils and major research funders such as the Wellcome Trust have been actively addressing these issues since the human genome sequencing projects started in earnest in 1997.

Policy Framework

Government Policy Initiatives

7. Following recent machinery of government changes, the main Government departments involved in developing and reviewing the policy framework for the research and development and implementation of genomic medicine into clinical practise are the Department of Health (DH), the Department of Innovation and Universities and Skills (DIUS) and the Department for Business Enterprise & Regulatory Reform (BERR).
8. DIUS funds initiatives through the Research Councils and the Technology Strategy Board, to encourage the development of the skills base and to encourage the development and trialling of potential medicines and innovative health technologies. DH is making rapid and substantial progress with implementing the Government's health research strategy *Best Research for Best Health*. The strategy will create a health research system, in which the NHS supports outstanding individuals working in world-class facilities, and in which leading-edge research focused on the needs of patients and the public is conducted.
9. DH has also set out a strategy for preparing the NHS to take advantage of genomic developments in the genetics White Paper published in 2003 and which has recently been reviewed (see paragraph 15).

Co-ordination of Research

10. BERR has a strong interest in genomic medicine because of its impact on the competitiveness of UK pharmaceutical and biotechnology industries. Companies in these sectors play important roles not only in generating new genetic knowledge but also in developing innovative technologies that will enable this knowledge to be taken up into routine healthcare. A particularly important initiative led by BERR (and previously DTI) in partnership with BBSRC and MRC was the LINK Applied Genomics collaborative R&D programme which involved 23 UK companies (21 SMEs) and 17 universities and research institutes. The programme aimed to use genomics to identify novel functionalities in biological systems for exploitation. There can be no doubt that the programme has been an outstanding success and has created significant value for the UK. Indeed, six of the companies involved in the programme have attracted some £500m further investments), and largely remain UK-based. BERR and DH jointly have, in partnership with the industry, undertaken three major initiatives - the Pharmaceutical Industry Competitive Task Force (PICTF), the Bioscience Innovation and Growth Team (BIGT) and the Healthcare Industries Task Force (HITF) which have helped advance genomic medicine in the UK, further information is available at paragraph 49 and **Annex A**.

11. DIUS provides funding of over £3 billion per annum to the seven Research Councils³. From this, the Research Councils fund a broad range of fundamental and translational research. Fundamental research seeks to understand basic mechanisms in health and disease and this research is conducted in universities and Research Council institutes. The research generates the underpinning knowledge that is essential for any product or technology development. Translational research is aimed at a range of application areas and much of it is conducted in collaboration with industry.

12. To develop the ideas and new potential drug targets identified by researchers, the Research Councils work closely with the Technology Strategy Board⁴ (TSB) as well as with the Health Departments in all four countries and agencies, National Institute for Health Research (NIHR) and industry. DIUS and the DH have jointly formed the Office for Strategic Coordination of Health Research (OSCHR). OSCHR's mission is to facilitate more efficient translation of health research into health and economic benefits in the UK through better coordination of health research and more coherent funding arrangements to support translation.

13. The TSB was established in July 2007 to invest in research and development; build partnerships between business, research and Government to address major societal challenges; and run a wide range of knowledge exchange programmes to help innovation flourish. With a leading role in UK innovation, it is envisaged that the TSB will help to build and maintain the country's global competitiveness. Over the next three years, the

³ <http://www.rcuk.ac.uk/default.htm>

⁴ www.innovateuk.org

TSB will develop and lead a strategic programme worth £1 billion through a number of key UK technology and application areas, to stimulate innovation in those areas which offer the greatest scope for boosting UK growth and productivity. The TSB is still developing the strategy for its future work, but its programme will be delivered in partnership with industry, other Government departments, Research Councils, Regional Development Agencies and the Devolved Administrations. The TSB is in discussion with MRC, DH and NIHR about co-ordinating research strategy, funding and areas for collaboration, further information is at **Annex A**. It will also liaise closely with OSCHR to discuss high-level research and technology policy issues.

14. The UK is one of the pioneers of genetic research, taking a lead role in the international Human Genome Project and its application to medicine. The Government remains committed to genetics research and it aims to make the United Kingdom the best place in the world for associated health research, development and innovation. A fundamental principle is that Government expenditure on research and development should result in more effective translation of health research into health and economic benefits in the UK and that it should deliver quality and value for money awarded contestably and transparently through rigorous competition. It will ensure, through the NIHR⁵, that necessary policies are in place and that resources are available to allow the NHS to contribute fully to this objective.

Integration of Genetics in the NHS

15. The Genetics White Paper set out the vision that the NHS should lead the world in taking maximum advantage of the application of the new genetic knowledge for the benefit of all patients. New genetic technologies are increasingly going to revolutionise the delivery of targeted health care and prevention of ill health. Over the past decade, the Government has set out a clear strategy for research into the link between genes and disease and to prepare the NHS to make maximum use of the new knowledge.

16. The White Paper *Our Inheritance, Our Future: Realising the potential of genetics in the NHS* (Cm5791-II)⁶ was presented to Parliament in June 2003. The main aim of the White Paper was to enable the NHS to make appropriate use of genetic knowledge and technology as it emerges. It announced £50 million of new expenditure to fund a broad-based programme of work including:

- strengthening specialised genetics services
- building genetics into mainstream services
- spreading knowledge across the NHS
- generating new knowledge and applications

⁵ <http://www.nihr.ac.uk/infrastructure.aspx>

⁶ http://www.dh.gov.uk/en/Publicationsandstatistics/Publications/PublicationsPolicyAndGuidance/DH_4006538

- ensuring public confidence

These commitments built on earlier investments to build capacity and capability in specialised genetics services.

17. The White Paper undertook to review this area after three years. The report of the review of the White Paper was recently published (copies have been supplied to the Committee). The review sought views from a wide range of scientific, professional and patient groups as well as Government committees with a responsibility for genetics. A summary of the progress on the commitments in the White Paper is at **Annex B**. It shows that significant progress is being made with key achievements as follows:

- Investment in specialist service capacity, in particular new laboratory technologies and training posts for laboratory scientists and genetic counsellors
- Support to develop the knowledge and understanding of the NHS workforce through establishment of the National Genetics Education and Development Centre (NGEDC)⁷ and the Genetics Conditions Specialist Library⁸ within the National Library for Health
- Funding for research in gene therapy, pharmacogenetics and health service issues related to genetics, which has included funding two novel gene therapy trials, the first of their kind in UK, for Duchene Muscular Dystrophy and childhood blindness and five year funding for the first NHS chair in Pharmacogenetics
- Support for service development pilots to trial new approaches to service delivery integrating genetics into other clinical specialty areas to provide more streamlined, patient centred, care

18. DH will continue to take forward developments to support both specialist genetic services and development of capability in the wider NHS, and to engage the public.

Advisory Framework

19. The Government has a comprehensive advisory framework to inform the development of policy relating to genomics. These include advisory Non-Departmental Public Body (NDPBs), professional bodies, advisory committees and *ad hoc* groups. This allows the monitoring of emerging scientific developments and consideration of their potential impact on clinical service. A list of some relevant bodies is at **Annex C**. Examples of their work are given in the appropriate sections of this memorandum.

⁷www.geneticseducation.nhs.uk/

⁸www.library.nhs.uk/genepool/

Horizon Scanning

20. DH, DIUS and BERR regularly undertake horizon scanning in order to have a mechanism to provide for the development of policies to raise future capability across Government, which will include that for genomic medicine. The current mechanisms of horizon scanning include:

- The National Horizon Scanning Centre Research Programme is part of the National Institute for Health Research (NIHR) and is funded by DH. The Centre investigates new and emerging health technologies that may require urgent evaluation and provides advance notice to the Department.
- The Horizon Scanning Centre⁹ in the Government Office for Science has been running a Wider Implications of Science and Technology¹⁰ (WIST) programme of stakeholder engagement to identify the safety, health, environmental, ethical, regulatory and social implications of new and emerging areas of science and technology. This has been closely integrated with a DIUS funded Sciencewise¹¹ programme (sciencehorizons¹²) of public engagement on emerging technologies.
- The Foresight Programme within the Government Office for Science uses robust evidence and rigorous futures methodologies to identify and understand the risks and opportunities that arise from uncertainty in order to improve strategy across Government
- A central Horizon Scanning Unit (HSU) and a distributed Horizon Scanning Network (HSN) have recently been established by DH to assist this integrated approach and to improve the capability for systematic and joined up futures thinking.

21. Further information on the Government Departments' horizon scanning capabilities are at **Annex D**

Genomics Research in the EU Framework Programme

22. The UK's approach to promoting genomic medicine was used during the negotiations of Framework Programme (FP) 7 to create funding modalities and topic areas that specifically addressed this policy area. The Commission is seeking to develop the genomics research funded in FP6 through its plans for FP7. FP7 will help integrate genomics, epidemiological, biological and biotechnology data and develop key technologies for health-related industries with a view to developing knowledge and capacity for intervention. This includes a 'high-throughput research' area aiming to catalyse progress in developing new research tools including fundamental genomics that will enhance significantly data generation and improve data and specimen (bio-

⁹ http://www.foresight.gov.uk/HORIZON_SCANNING_CENTRE/index.html

¹⁰ http://www.foresight.gov.uk/HORIZON_SCANNING_CENTRE/WIST/Index.html

¹¹ <http://www.sciencewise.org.uk/>

¹² http://www.sciencewise.org.uk/html/projects.php?source=projectdetail&project_ID=9

banks) standardisation, acquisition and analysis. There is also a focus on 'Large-scale data gathering' to use high-throughput technologies to generate data for elucidating the function of genes and gene products and their interactions in complex networks in important biological processes.

23. Genomics research will also be included in the Innovative Medicines Initiative (IMI), which is a public private partnership to be funded over the next 10 years using €1bn from the European Commission (via the FP7 Health Theme) and €1bn in kind contributions from industry. The aim of the IMI is to support the faster discovery and development of better medicines and to enhance Europe's competitiveness by ensuring that its biopharmaceutical sector remains a dynamic high-technology sector. The knowledge management strand of IMI will aim to maximise on technologies such as genomics by providing platforms to analyse large amounts of information in an integrated and predictive way. Genomics and pharmacogenomics will be included in the IMI strands aiming at improving predictivity in drug safety and drug efficacy evaluation.

24. EuroGentest¹³ is an EU-funded Network of Excellence (NoE) which looks at all aspects of genetic testing such as, Quality Management, Information Databases, Public Health, New Technologies and Education. Through a series of initiatives, EuroGentest encourages the harmonization of standards and practice in all these areas throughout the EU.

Regulation

25. The Medicines and Healthcare products Regulatory Agency (MHRA) is the Competent Authority for medical devices in the UK. All genetic test kits, whether they are regarded as new technology or not, which detect a gene from blood or other human fluid samples *in vitro* are regulated under the In Vitro Medical Devices (IVD) Directive which has been transposed into UK law by the Medical Devices Regulations 2002, further information is provided in **Annex E**.

26. The Human Fertilisation and Embryology Authority¹⁴ (HFEA) was created by the Human Fertilisation and Embryology Act 1990. The first body of its kind in the world, the HFEA was established to regulate fertility treatments involving *in-vitro* fertilisation (IVF), the use of donor gametes (eggs and sperm) and embryos, the storage of gametes and embryos and research involving human embryos. The HFEA currently licences the testing of embryos, created by *in vitro* fertilisation, for the presence of inheritable genetic conditions, in certain circumstances.

Ethical and Societal considerations

27. One of the societal or economic Grand Challenges that the Research Councils announced in December 2007 will address lifelong health and well-

¹³ <http://www.eurogentest.org/>

¹⁴ <http://www.hfea.gov.uk/>

being, in response to the Treasury's Grand Challenge on the effects of an ageing population on economic competitiveness. The Research Councils' Grand Challenges will be developed in conjunction with researchers, users and the public and will include basic research through to application plus studies on risk governance, economics, and social implications.

28. The Human Genetic Commission¹⁵ is the UK Government's advisory body on new developments in human genetics and how they impact on individual lives. It advises the Government on human genetics with a particular focus on the social, ethical and legal issues. The HGC reports to Health and Science Ministers and Ministers in Scotland, Wales and Northern Ireland. Its terms of reference include a role in advising on strategic priorities for research and the delivery of genetics services by the NHS. Recent discussions by the HGC have included research priorities in genetics; the protection of genetic information, such as in research or clinical databases; the use of genetic knowledge in reproductive decision-making and the use of genetic information for non-clinical purposes, such as insurance, employment and forensics. The Government is currently considering the recommendations of an independent review of the HGC which has endorsed the important contribution the committee makes.

29. The Patient Information Advisory Group (PIAG)¹⁶ was established to provide advice on issues related to the use of patient information and to advise on powers under Section 251 of the NHS Act 2006, which lifts the common law duty of confidentiality in limited circumstances. The membership of PIAG is independently appointed and includes patient, lay members, healthcare professionals, and researchers. Its remit includes both data based research and the identification of potential research subjects.

30. It should be noted that the Health Bill currently passing through Parliament proposes to transfer PIAG's functions to a newly established National Information Governance Board (NIGB) and provides this new body with additional powers.

31. In 2006, the government's health research strategy *Best Research for Best Health* announced that the UK Clinical Research Collaboration, DH R&D and NHS Connecting for Health, would work closely to ensure that the data collected via the NHS Care Record Service and supporting infrastructure meet the needs of researchers and public health practitioners, strictly within the bounds of patient confidentiality. The UKCRC formed an expert advisory group, and published recommendations in mid 2007. The Department responded by establishing a Research Capability Programme in NHS Connecting for Health. Over the next few months, the programme will complete its enabling phase, which is defining the major elements to be delivered over the next few years.

¹⁵ <http://www.hgc.gov.uk/Client/index.asp?ContentId=1>

¹⁶ www.advisorybodies.doh.gov.uk/PIAG

32. The policy framework for research uses of genomic information is based on a careful analysis of the legal, social and ethical implications. Research in the NHS is conducted within a framework of research governance policies based on the law, international best practice and on a number of international consensus statements, such as the World Medical Association's Declaration of Helsinki. In 2005, the Department reissued the *Research Governance Framework for Health and Social Care* as a core standard¹⁷ for health care organisations. The Department's guidance on the system of research ethics committees¹⁸ is under review following the creation of a National Research Ethics Service. The NRES is implementing a programme of measures¹⁹ to harmonise and streamline the operation of research ethics committees.

33. The Council of Europe's Steering Committee on Bioethics (CDBI) provides an international forum focussing on the ethical and legal implications of genetics, and DH officials represent the UK. A Protocol to the Convention on Human Rights and Biomedicine covering genetics has been under development and is currently awaiting approval by the Council of Europe. This expands on the core principles of the parent Convention covering matters such as consent and privacy. The Council of Europe is also considering the need for further work on pre-implantation genetic testing, prenatal genetics testing and genetics and insurance.

Research and Scientific Development

34. The pace with which new genetic knowledge is generated is increasing. Better DNA sequencing strategies, perhaps dependent on identifying genetic differences between different individuals rather than determining entire genomic sequences (the absolute similarity between genomes from separate individuals exceeds 99%) will alone transform the availability of genetic information on an individual. Improved algorithms for assessing the interactions between an individual's genes, environment and lifestyle will result in the development of an increasing number of prediction and prevention strategies. The benefits of some of these will be easy to identify and justify uptake into clinical practice. However, this will not be the case for all and this emphasises the need for ongoing research and development into the ethical, legal and sociological frameworks needed to support the adoption of new genetic information in healthcare.

35. In 2003 the DH invested over £15 million in three specific programmes of research on genetics based health services, pharmacogenetics and gene

¹⁷ *Department of Health Research Governance Framework for Health and Social Care*, 2nd edition, DH April 2005, issued as a core standard for health care organisations. Health care organisations have to take these standards into account in discharging their duty of quality under Section 45 of the Health and Social Care (Community Health and Standards) Act 2003.,

¹⁸ *Governance Arrangements for NHS Research Ethics Committee*, DH 2001

¹⁹ *Building on Improvement – Implementing the recommendations of the Report of the Ad Hoc Advisory Group on the Operation of NHS Research Ethics Committees*, National Patient Safety Agency, August 2006

therapy in order to generate new genetics knowledge relevant to healthcare. This formed part of the £50 million funding package announced in the genetics White Paper.

36. The programme on genetics based health services was aimed at consolidating and developing a robust evidence base to inform the policy, planning and implementation of health care services with a genetic element. It was concerned with the commissioning, organisation, management and delivery of services and with aspects of patient, public and societal attitudes and behaviour that may influence the design or delivery of care. Seven projects were funded, the majority of which were designed to investigate ways to better communicate both genetic information and genetic risk to different population groups. These projects are all ongoing and an assessment of the impact of any outputs is therefore not possible at the present time however, it is already clear that they will have some influence on the shape of future policy development.

37. The programme on pharmacogenetics was specifically focused on existing medicines that are commonly used or that have a reduced benefit due to possible genetic-related toxicity. This focus was created with the recognition that the pharmaceutical and biotechnology industries were investing heavily in pharmacogenetic research on the new medicines that are to be marketed in the future. Six projects were funded but, perhaps more importantly, funding was also provided to establish the first NHS Chair of Pharmacogenetics and a supporting research team at University of Liverpool. It is intended that this Chair should increase research capacity and knowledge in this important field and facilitate the development of a network of researchers across the NHS capable of evaluating and then introducing pharmacogenetic tests into the routine prescription of medicines.

38. The programme on gene therapy was made up of two components. First, research into the long-term safety of the use of gene therapy vectors that are designed to insert therapeutic genes into human genetic material. The object here was to initiate the development of an evidence base that would minimise the risks and maximise the benefits of gene therapy. Four projects were supported; evaluating the outcomes and impact of this research is still on-going.

39. Second, research into the development of gene therapy to treat single gene disorders. This was designed to stimulate research in an area that has potential benefit to a significant proportion of the UK population (estimated at over three-quarters of a million) and that is unattractive to commercial investment because of the associated high risks and low financial returns. The budget for this part of the gene therapy research programme was supplemented to enable clinical researchers to gain access to gene therapy vector production facilities capable of operating to the exacting standards of good manufacturing procedures. Investment in this research has led to clinical trials for assessing treatments for severe combined immunodeficiency (SCID) in children, a form of childhood blindness, Duchenne muscular dystrophy in boys and myeloma. Two additional trials

for the treatment of leukaemia and cystic fibrosis are also planned. A clinical trial for the treatment of cystic fibrosis is being planned.

40. Supporting these specific research programmes in harnessing the potential of human genetics to improve healthcare was the network of UK Genetics Knowledge Parks (GKPs) announced in 2002. This network was established as a joint initiative between the Department of Health, what was then the Department of Trade and Industry and the National Assembly for Wales. Six GKPs were created in Cambridge, Cardiff, London, Newcastle, Oxford and the North West. Together they investigated a diverse array of topics, which encompassed the translation of new science into diagnostic, testing, screening and treatment pathways, a consideration of the complex ethical, social and legal issues involved in this process and the need for professional education and public engagement.

41. The GKPs did excellent pioneering work and although the dedicated funding for the English (but not Welsh) Genetics Parks has come to an end the work they began is continuing within the separate institutions and wider networks. They have made a significant impact in alerting the NHS to some of the issues needed to embed genetics into mainstream medicine and service delivery. They have done this by creating multi-disciplinary environments where clinicians and laboratory workers can meet with teachers, lawyers, politicians, ethicists industrialists, patient groups and the general public to explore the ways in which genetic technologies can best be deployed in health care settings. They have demonstrated that the translation of genetic research into clinical practice is a complex process that is as reliant on organisational structure as scientific and technological developments. The work of the Oxford GKP played a significant role in the creation of one of the new Biomedical Research Centres (see later).

42. Progress in implementing the recommendations made in the Genetics White Paper has been reviewed recently (Annex B) and the feedback obtained during this process identified the related disciplines of proteomics and transcriptomics as additional key areas for consideration. It was felt that research on these topics was essential for further understanding the mechanisms that underlie disease and that this was likely to lead to new and effective treatments. In addition, further epidemiological research was considered essential to understand the significance of new discoveries about the existence and effects of polymorphisms (common variations in genetic make-up).

43. The outcomes of this research will improve our understanding of the influence of genetic variations on the risk of common chronic conditions. This will become increasingly important to people making lifestyle choices to reduce their risk of these diseases. This underlines the importance of health professionals having better access to both information about genetic conditions and management support to incorporate genetics advances into their practices.

44. The genetics research programmes were time limited. Continuing and future support from the DH for research in this area will come from the Department's mainstream funding for health research. The Department's research and development budget for the NHS for 2007-08 is £733 million - 7.7 per cent higher in real terms than in 2006-07. This pattern of increases in Government funding for health research is set to continue across the current comprehensive spending review period. It will lead to a budget of over £1 billion for DH and a total Government investment in health research, including funding for the Medical Research Council (MRC), of over £1.7 billion per annum by 2010-11.
45. The DH established the National Institute for Health Research (NIHR) in April 2006 to deliver the objectives set out in *Best Research for Best Health*. The NIHR is making a substantial investment in the infrastructure to support experimental medicine in the NHS, through its support for Biomedical Research Centres and Units and Clinical Research Facilities, the majority of which are undertaking research relevant to genomic medicine
46. Genome technologies are employed extensively within the first eleven NIHR Biomedical Research Centres, which receive substantial levels of funding (£468m over five years) to translate fundamental biomedical research into clinical research that benefits patients. These Centres are the most outstanding NHS/University partnerships in the country and are leaders in scientific translation and early adopters of new insights in technologies, techniques and treatments for improving health. The Biomedical Research Centre in Manchester, announced on 19 March 2008, will be exclusively concerned with the application of genetic technologies to diagnostic procedures and treatment in a number of key areas of healthcare. It will build on the work of the former North West GKP.
47. The NIHR is also establishing 12-16 Biomedical Research Units in 2008-09 to undertake translational clinical research in the following priority areas of high disease burden and clinical need, which are under-represented in the existing NIHR Biomedical Research Centres: cardiovascular disease; deafness and hearing problems; gastrointestinal (including liver) disease; musculoskeletal disease; nutrition, diet and lifestyle (including obesity); and respiratory disease. Again, genomic technologies will be fundamental to the research undertaken by these Units, each of which will receive at least £3.75m over four years
48. Finally, a fellowship fund has allowed young health professionals to travel abroad with the aim of bringing back to the NHS relevant skills in genetics healthcare, research and new laboratory technology. In addition, eminent genetics scientists have visited the UK and contributed to genetics policy development and public health research programmes.

Translation

49. The Pharmaceutical Industry Competitive Task Force (PICTF) process set a new direction of travel in relation to the relationship the Government had

with the pharmaceutical industry as it provided a structured, action-oriented platform for effective dialogue. It was recommended in the PICTF report (2001) that this close working should continue through the Ministerial Industry Strategy Group (MISG). MISG is a high-level group co-chaired by the relevant DH Minister and a company Chief Executive and its role is to take a strategic overview of the UK environment.

50. Bioscience Innovation and Growth Team (BIGT) concluded that that the bioscience-related industries would play a major role in:

- population screening to identify the proportion of the population affected by a specific condition or disease
- research to elucidate the underlying genetic and cellular mechanisms of the disease
- disease detection
- research to elucidate which cellular targets may be most productive for drug development
- identification of subpopulations to be tested in clinical trials of drugs in order to claim targeted clinical efficacy only in the relevant patient group
- screening prior to prescribing treatment to identify patients who would benefit from the drug in question

51. A significant outcome of the BIGT process was the creation of the National Clinical Trials Agency (NCTA), which supports excellence in clinical trials and clinical research within the NHS. However, the overall effect of the BIGT initiative is to be reviewed and refreshed through a review to be chaired by Sir David Cooksey to determine what more needs to be done to drive the development of the industry and to ensure its global competitiveness.

52. There will be three areas of work a) a stocktake workstream – reviewing progress against the Bioscience 2015 recommendations b) a finance and investment workstream – looking at the particular issues faced by the sector in raising finance and investment and c) a new ideas workstream –looking at emerging issues impacting on the competitiveness and future of the medical bioscience sector.

53. Finally, in recognition of the key role played by medical devices and diagnostics in the delivery of high quality health and social care services, the Healthcare Industries Task Force (HITF) was established in 2003. This resulted in the creation of the HITF Strategic Implementation Group (SIG) to oversee the start of the delivery of HITF outputs, and to continue to develop the dialogue between Government and the industry. A key issue continued to be how to balance the NHS's need to secure good value for money with investment in innovation where the returns are likely to be realised only in the longer term.

54. These three initiatives (PICTF, BIGT, HITF) collectively, but sometimes implicitly, recognised that genomics is enabling industry

- To identify which patients will react best to drugs and conversely to detect genotypes which may be liable to have an adverse reaction to particular drugs. Blood samples are routinely taken and phenotype and genotype assessed in selecting patient cohorts for clinical trials and thereby select-in patients where the drug is most likely to work. For existing drugs diagnostic kits can be used to assess genotype and customise the dose of the drug that is given
- To diagnose diseases much more precisely and treated with highly specific therapies- a move towards personalised medicines. Examples of this might include gene therapy and RNA interference-based drugs.
- To develop diagnostic assays which will detect biomarkers of disease at onset and progression while in disease management to use such biomarkers as surrogate indications of disease in developing drugs
- To develop new therapeutics and vaccines given the new molecular targets (both in humans and in pathogens) identified by genomics

55. This, in turn, has facilitated significant cross-sector collaboration in the following ways:

- Genomics has led to a surge in the amount of data being created and that needs to be analysed. Collaborations with the science base and with technology providers in areas such as data mining are widespread. The MRC, Wellcome Trust and the Sanger Institute are actively involved in such studies
- The Bio-pharmaceutical Industry collaborates extensively on pre-competitive research with the research councils in the UK, with many Universities outside of the UK and the National Institute of Health (NIH) in the US. There is extensive collaboration within the sector and with enabling technology supply companies

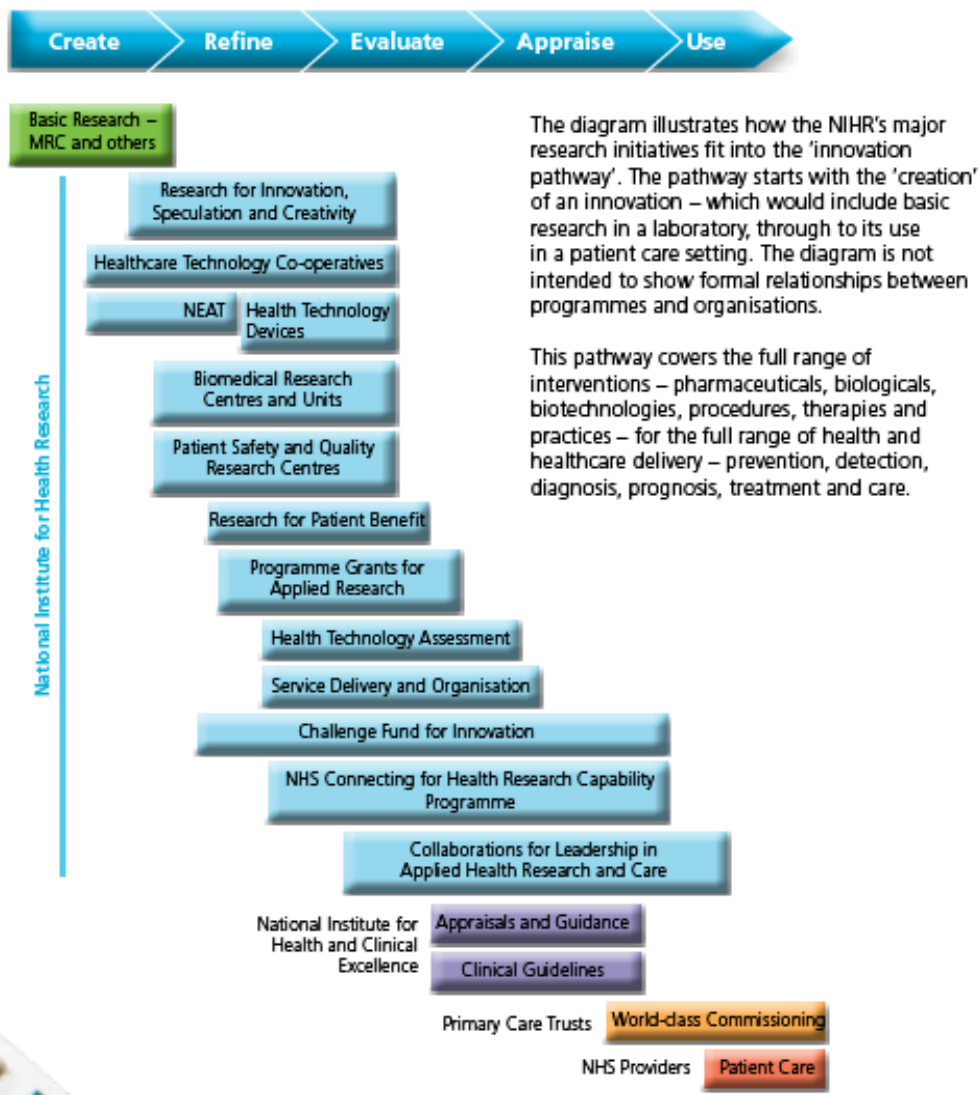
56. Recent developments in NIHR funding for translation and innovation:

- One of the key points made in the Cooksey Review was the need to bridge the gap between basic scientific discoveries and new ways to prevent, diagnose and treat disease. To bridge this gap we are working closely with the MRC under the umbrella of the newly established Office for Strategic Coordination of Health Research (OSCHR) Board to develop an integrated strategy for publicly funded health research.
- In November 2007, in partnership with the MRC, DH announced a new raft of measures for clinical trials designed to help turn research discoveries into benefits for patients faster:

- The Efficacy and Mechanism Evaluation (EME) Programme will help to move promising technologies forward by evaluating their efficacy and safety by assessing their clinical and cost-effectiveness. The programme, funded by the MRC, will start in April 2008.
 - The Patient Research Cohorts Initiative will identify small, carefully defined groups of patients with particular illnesses, in order to throw light on previously poorly understood aspects of how these illnesses progress.
 - The Methodology Research Programme will support the development of new, improved systems and theories for health research, because research is only able to provide reliable and relevant evidence if it is carried out using the most powerful and appropriate research methods. The programme will investigate the tools, theories and disciplines that underpin the design, analysis and evaluation of research in the health sciences.
- These new initiatives will complement existing NIHR's research programmes²⁰, which are illustrated in the 'innovation pathway' diagram.

²⁰ www.nihr.ac.uk

The central role of NIHR research in the innovation pathway



Diagnostics industry

57. The medical devices industry covers a diverse range of products, including medical commodities, hi-tech hospital equipment, robotic implements and *in vitro* diagnostic devices. In the latter case, there is considerable potential for their application in relation to :

- population screening to identify people at a higher risk of a disease or its complications
- research to elucidate the underlying genetic and cellular mechanisms of the disease
- disease detection
- research to elucidate which cellular targets may be most productive for drug development

- identification of subpopulations to be tested in clinical trials of drugs in order to claim targeted clinical efficacy only in the relevant patient group
- screening prior to prescribing treatment to identify patients who would benefit from the drug in question

Innovation

58. It is recognised that, while Britain is a leader in basic science and the development of health innovative techniques and technologies, adoption of these innovations is variable across the health and social care systems. The Health Innovation Council (HIC)²¹ has been established to promote the benefits of innovation to the NHS and social care and provide leadership in supporting innovation from discovery through to adoption. It will provide leadership and advocacy with key decision-makers in the NHS on the benefits to patients, the NHS and the country of adopting cost-effective new technologies and models of care.

59. The National Innovation Centre²² (NIC) was set up as a cross-governmental and HITF recommendation to assist innovators who may have early innovations, navigate the innovation landscape. The NIC is not focused on genomics but sees examples of early stage technologies including, medical devices, diagnostics and training packages. The NIC facilitates and helps smooth the path to broader uptake by interaction with other agencies on the landscape. The NIC also coordinates the local Innovation Hubs²³.

60. The nine local Innovation Hubs manage intellectual property (IP) emanating from NHS staff at the local level. This IP may include any research from NHS staff and might include genomic/genetics research.

Implications for the use of genetic information

61. The Government has taken careful note of the ethical, legal and social implications of the generation and storage of genomic information. DH and the then Department of Trade & Industry (with the Research Councils and the Wellcome Trust) were closely involved in the discussion about the establishment of large infrastructure projects like the UK Biobank.

62. The 2003 Genetics White Paper noted the concerns are about breaches of privacy and confidentiality (for example, inappropriate sharing of sensitive medical information) leading to discriminatory treatment or stigmatisation. It re-affirmed the role of the Human Genetics Commission and the Genetics and Insurance Committee in addressing the potential for the abuse of genetic information in employment and insurance.

²¹

http://www.dh.gov.uk/en/Managingyourorganisation/Commissioning/Worldclasscommissioning/Innovation/index.htm?IdcService=GET_FILE&dID=151015&Rendition=Web.

²² www.nic.nhs.uk

²³ www.nic.nhs.uk/About/InnovationsHubs/Pages/Introduction.aspx

63. In 2006 the Human Genetics Commission (HGC) concluded, following a survey of employment and genetic organisations, that there was no significant evidence of genetic testing occurring in the workplace. There remains, however, concern that genetic information may be misinterpreted or given improper relevance in an employment context, or that employees might be induced or pressurised to reveal results of genetic tests already taken.

64. The use of genetic test information by employers is covered in Part 4 of the Information Commissioner's statutory Employment Practice Data Protection Code. This sets out in detail the requirements of the Data Protection Act 1998 and incorporates many of the restrictions on the use of genetic information that were recommended by the HGC. It considers that genetic testing is of uncertain predictive value for most employment contexts and use of genetic test results should only be introduced after careful consideration, if at all.

Insurance

65. In light of concerns that people could be discouraged from taking medically important genetic tests that then made insurance unobtainable or unaffordable, the Government and the ABI (acting on behalf of the insurance industry) have agreed a number of safeguards.

- GAIC was set up in 1998 to evaluate applications from the insurance industry for the use of results from predictive genetic tests in setting insurance premiums. It is a tripartite body reporting to Health, Treasury and DIUS Ministers with the Secretariat provided by DH.
- In 2001, a 5 year moratorium on the use of such test results was announced by the ABI in response to a critical House of Commons Select Committee report. The Moratorium was extended to 2011 and incorporated into a Concordat on Genetics and Insurance²⁴ agreed between the Government and the ABI, which was published in March 2005.

66. Under this Concordat and Moratorium, insurance companies can only ask for the results of predictive genetic tests if (a) the test has been approved by GAIC and (b) the policy is for more than £500k of life cover or £300k for other types of health insurance (critical illness, income protection and long term care). To date, only one predictive genetic test has been approved by GAIC in 2000 which is for Huntington's Disease for life insurance policies over £500k. There will be an operational review of the Concordat in 2008 to ensure that the Concordat remains up to date and reflects current arrangements for oversight of the use of genetic test results in insurance.

²⁴

http://www.dh.gov.uk/en/Publicationsandstatistics/Publications/PublicationsPolicyAndGuidance/DH_4105905).

67. The current policy forms the basis for the position in the 2007 consultation²⁵ which set out the Government's proposals for a Single Equality Bill for Great Britain. The Government's position was that "there is no current justification for legislating to prohibit genetic predisposition discrimination" given the safeguards in the Concordat. The response to the consultation setting out the Government's position is likely to be published in the summer.

Direct to consumer genetic testing

68. A number of bodies, including the Human Genetics Commission, and Sense about Science, have raised concerns about the provision of genetic and similar tests directly to the public.

69. Under EU legislation, the MHRA does not have the legal basis to grant pre-market approval or otherwise control the activities of those providing genetic test services direct to the public. At present, a manufacturer of these genetic tests self-certifies compliance with the IVDD. The Notified Body's role is restricted to higher-risk tests and to the self-test element, including information provided to the user to enable him to use the kit properly and interpret the results correctly. The IVDD also does not contain any provisions relating to controls on advertising and promotion of genetic testing services so this also comes outside the scope of MHRA's regulatory responsibility. Further information is at **Annex D**

70. In view of this, the Government has welcomed the HGC's proposal to work with relevant stakeholders to develop a comprehensive code of practice or guidelines, based on the findings of the HGC's previous reports on genetic testing services provided directly to the public. This will include working with the relevant agencies, such as the MHRA, the Human Tissue Authority, the Advertising Standards Agency and OfCom. At this early stage of development of the market for such testing services, there are advantages to having a self-regulatory system to ensure that any new developments can be accommodated flexibly without undue delay.

UK Biobank

71. There is intensive research underway to use the results of the Human Genome Project to map genetic differences in individuals to show variations associated with common diseases like cancer, stroke, diabetes, heart disease and dementia. This is using some of the unrivalled collection of patient samples and data in the UK.

72. UK Biobank²⁶ which is jointly funded by DH, MRC and the Wellcome Trust, is developing a prospective resource of 500,000 people aged 40-69 from

²⁵ Discrimination Law Review: A Framework for Fairness: Proposals for a Single Equality Bill for Great Britain

<http://www.communities.gov.uk/publications/communities/frameworkforfairnessconsultation>

²⁶ www.biobank.ac.uk

around the UK, involving extensive baseline information, physical measures, biological samples and medical information. Generation Scotland (like many other studies) is complementary to UK Biobank and will help to develop the knowledge of genes which contribute to health or ill-health. Such studies aim to help researchers better understand the causes of disease and to find new ways to prevent and treat different conditions. Understanding the genetic basis of disease is one component of building a full picture of health processes. UK Biobank and other biobanks provide the opportunity to investigate how genetic factors combine with lifestyle and other factors to cause disease.

73. UK Biobank will build on existing genomic information, provide further opportunities for genomic investigation and discovery, and, importantly, become increasingly valuable (and cost effective) to researchers to extend studies into the assessment of the complex interplay between the effects of different factors (e.g. genetic and environmental) in the development of, and recovery from, disease. The suite of samples stored will support study of a wide range of biomolecules and will allow many types of assays (e.g. proteomic and metabolomic) to be undertaken in addition to genetic analyses. Studies of the scale and complexity of UK Biobank will be unique in having the power to address these research questions which build on the outputs of genomic research data. These complementary approaches will provide a treasure trove of information that will transform health research and treatment in the years to come.

Genome-wide association studies

74. Biomarkers are characteristics that are objectively measured and evaluated as indicators of biological and pathogenic processes or pharmacological responses to a therapeutic intervention. They play a key role in understanding the aetiology and progression of disease, as well as the mechanism of action of therapeutic agents and investigating the response of patients to treatment. Biomarkers are an essential part of the drug discovery process, being used at every stage from target identification to patient stratification.

75. Genome-wide association studies provide a comprehensive approach to identification of common genetic variants (one type of “biomarker”) across a population which are either directly associated with the risk of disease (e.g. cancer risk) or influence the inheritance of commonly measured quantitative traits, (another kind of biomarker e.g. levels of cholesterol as a risk factor for cardiovascular disease). These studies will lead to the discovery of new genetic factors and disease causing molecular pathways that will open up new therapeutic avenues.

76. Genome-wide association scans will also help screen for biomarkers associated with the clinical response to treatment and may lead to the identification of single nucleotide polymorphisms (SNPs) and differentially expressed genes that are related to and predictive of the responder status. The use of genomics and gene expression platforms at the level of the entire

genome therefore offers unique opportunities for biomarker research and its translation to patient benefit in terms of tailoring treatments for optimal efficacy and safety according to individual's genetic make-up.

77. The related use of gene chips (microarrays) will make it possible to predict the response of cancers to chemotherapy, diagnose complex syndromes causing mental impairment in children and screen people to target prevention strategies for diseases like cancer or diabetes. The capacity of chips and other high throughput screening devices is rapidly developing – it is now possible to analyse 500,000 variations (SNPs) per chip. New evidence is also coming out that many conditions result from duplications, deletions or inversion of small amounts of the chromosome. These copy number variations are increasingly associated with complex conditions such as autism.

78. However it is important to manage public expectation on the predictive capacity of these genetic variations. The interpretation of these gene associations with disease risk is complex as many common chronic diseases are caused by an interplay of genetic and environmental factors. Large scale, long-term population studies will add to our understanding of these influences. The communication of genetic based risk to individuals is a highly skilled task that has considerable resource implications for health services. More integrated research is needed before clinicians can use this information to advise individuals on their personal risk. Moreover, there is conflicting evidence as to whether knowledge of gene- based personalised risk for chronic disease such as cancer or CHD actually leads to the effective modification of risk factors for those individuals.

Use of genomic information in a healthcare setting

79. The 2003 genetics White Paper recognised that over time the relevance of genetics across other medical specialties would become increasingly clear to clinicians and others working outside the genetics specialty. Specialist genetics services will continue to play a leading role in diffusing new genetics advances across the spectrum of medical care. It will also be essential to encourage the take-up of genetics technology through service development so that other clinical services can explore ways of using genetics knowledge to help patients with inherited conditions and their families.

80. The White Paper identified a need for central support to encourage the development of new patient pathways and types of service in areas of mainstream service planning. . DH has funded a number of initiatives to pilot ways of integrating genetics knowledge into other clinical specialties (see **Annex B**) for example the joint DH and Macmillan cancer genetics pilots partnership programme (co-funded with Macmillan Cancer Support) which have worked to integrate a more efficient and accurate risk assessment for families into cancer services. This has supported better quality patient

centred care and more efficient use of NHS resources by ensuring patients are seen by the relevant part of the service.

81. Two bodies, the UK Genetic Testing Network (UK GTN) and the Genetics Commissioning Advisory Group (GenCAG) play an important role in supporting and advising the NHS on the role of genetics in clinical care and the development of services.

82. UK Genetic Testing Network (UK GTN)²⁷ is a collaborative group of NHS laboratories, clinical genetics specialists, patients' representatives and service commissioners. Its work is overseen by the UK GTN Steering Group and it uses a series of working groups to provide advice and information to the NHS on service developments. The steering group reports to the Genetics Commissioning Advisory Group (GenCAG). One of its key achievements has been the development of a rigorous evidence-based system for introducing new tests into NHS service that assesses both scientific validity and clinical utility. This service has provided NHS service commissioners with valuable evidence about the clinical usefulness of new tests to help inform decisions about what tests to fund. The UK GTN also maintains a website, which holds comprehensive and annually updated information on all molecular genetic tests available through the NHS in the UK, together with details of laboratories in the UK that provide each test.

83. GenCAG was set up to provide advice to commissioners of genetics services to enable them to provide appropriate services for NHS patients and their families. Its terms of reference are:

- To take a strategic national overview of genetics in healthcare delivery
- To provide advice to NSCAG (now the National Commissioning Group (NCG) and to Specialised Commissioning Groups (SCGs) in delivering appropriate services (within available resources) to NHS patients and their families who require investigation, diagnosis and counselling for conditions with a genetic basis or where genetic technologies underpin service provision
- To enable NHS patients and their families to benefit from advances in genetic technologies shown to be effective and to have clinical utility.

84. The UK National Screening Committee (UK NSC) advises Ministers and the NHS in all four UK countries about all aspects of screening policy and supports implementation. Using research evidence, pilot programmes and economic evaluation, it assesses the evidence for programmes against a set of internationally recognised criteria. These criteria cover the condition, the test, the treatment options and effectiveness and acceptability of the screening programme. Assessing programmes in this way is intended to ensure that they do more good than harm at a reasonable cost. The criteria for national screening programmes have been expanded to take account of

²⁷ www.ukgtn.nhs.uk/gtn

family issues and consent processes when the condition that is being screened is genetically determined.

85. Genetics and inherited disease feature in a number of population screening programmes, including antenatal screening for Down's syndrome, sickle cell disease, thalassaemia, cystic fibrosis and Medium Chain Acyl CoA Dehydrogenase Deficiency (MCADD).

Education

86. Advances in genomic medicine will require clinical teams to be knowledgeable about the implications of genetic tests in the management of patients. The genetics White Paper established the National Genetics Education and Genetics Centre (NGEDC) in Birmingham. The Centre will consider the genetics educational needs of health professionals who are not genetics specialists and to work with the relevant professional and regulatory bodies to get genetics incorporated into curricula and continuing professional development to meet those needs. For example the NGEDC has developed with Skills for Health a competence framework for health professionals working outside specialist genetics departments. It is also working with the medical Royal Colleges to incorporate genetics into the new curricula for junior doctors' training.

87. The White Paper also established a genetics portal in the National Library for Health, which allows health professionals to access information about genetics and clinical practice from the workplace, the Genetics Conditions Specialist Library²⁸.

Regulation of Advice

88. Within the NHS advice about genetics testing is covered by the same principles as other areas of medical advice and consent. The professional bodies represented in the British Society of Human Genetics provide advice and guidance on ensuring that genetic information is communicated in an appropriate manner. More generally, DH has made it clear that patients have a fundamental legal and ethical right to determine what happens to their own bodies. Valid consent to treatment is therefore central to all forms of healthcare, from providing personal care to undertaking major surgery. This also requires the provision of appropriate information and ensuring that the patient understands that information and is has the capacity to take the decision.

Training and Workforce issues

89. The Chief Scientific Officer in DH is currently leading a programme to modernise healthcare scientist careers (Modernising Scientific Careers) to develop a scientist workforce that is trained and supported to adapt to the changing demands of a NHS service where technologies and

²⁸ www.library.nhs.uk/genepool/

methodological approaches to work will change with increasing frequency. The programme aims to reduce the time taken to become a registered geneticist from four to three years, will expose those in training to a broader range of diagnostic modalities and will offer a specific outcome based higher specialist training programme.

90. Workforce planning is a matter for local NHS organisations to deal with, as they are best placed to assess the health needs of their local health community and to commission the required number of education and training places to meet those needs. As part of this approach, Strategic Health Authorities are expected to be satisfied that local workforce plans are sufficiently robust to deliver the planned activity. DH recognises the importance of good workforce planning and we work closely with the SHAs to ensure these robust planning arrangements are in place. A key support to SHAs in this is the Workforce Review Team (WRT) , a national body working on behalf of the NHS in England. WRT's role is to review in specific detail, the supply and demand of the healthcare workforce, and advise on the most practical and effective use of resources. Working in collaboration with other health and social care organisations, WRT draws its information from a wide range of sources about the workforce needed to deliver high quality modern health and social care, and to meet changing demands

91. Following the Health Select Committee report in March 2007, DH accepted there was a need to review workforce planning in the NHS. That is why as part of Lord Darzi's NHS Next Stage Review, DH is working with all stakeholders (including the Unions and Royal Colleges) to overhaul our approach to workforce planning, education and training in the NHS.

Conclusion

92. The Government remains committed to:

- the translation of knowledge of human genetics into clinical practice to improve the healthcare pathways involved in the prevention, diagnosis and treatment of disease.
- developing the genetics capacity and capability within the NHS
- ensuring that there is a world-class environment for conducting and using NHS health research

93. We will assist the Committee in providing any further information that is required and look forward to the outcome of the Committee's deliberations.

Government/Industry Initiatives

Technology Strategy Board

1. The Technology Strategy Board has a role to ensure UK business is successfully competing at the forefront of technology and innovation developments globally. This includes providing a longer term perspective of future technology and innovation needs. With a focus on technology enabled innovation, the TSB will understand the support required by business to make a difference and provide a coherent view of the support available, both from the TSB and from others, enabling easy access to the most relevant support.
2. The Technology Strategy Board considers that two priority areas for UK Healthcare innovation are –
 - Data Management: harnessing the vast amounts of data generated from high throughput technologies requires innovative and intelligent data mining and translation technologies to create the next generation of therapies; and
 - Translational Medicine: translating the basic discoveries into therapies for patients requires a multidisciplinary approach tying together the above disciplines with clinical research and development. Ensuring a joined up approach between the different disciplines requires effective co-ordination of information and knowledge between the different research areas.
3. To date, the Technology Strategy Board has funded several collaborative (between academia and industry) R&D projects focussed on biomarker discovery and translation. It is also funding a further two initiatives that are relevant, but not directly related, to genomics.
 - Stem Cells for Safer Medicines. This is a public-private collaboration between five public funders, (DH, DIUS, MRC, BBSRC, Scottish Executive) and three multinational pharmaceutical companies (Astra-Zeneca, GSK and Hoffman La Roche) and facilitated by TSB and the Association of British Pharmaceutical Industries. It will enable the creation of a bank of stem cells lines with open protocols and standardised systems in stem cell technology that will enable consistent differentiation of stem cells into stable homogenous populations of particular cell types, with physiologically relevant phenotypes suitable for toxicology testing in high throughput platforms; and
 - A pilot Healthcare Technology Co-operative. This will bring patients and carers, academics, clinicians, nurses and other healthcare providers together with industry to catalyse the development of new healthcare products that will empower people

with debilitating conditions and enhance their lives. The pilots will be used to assess the benefits and effectiveness of this kind of partnership working. They are a result of collaboration between the DH, DIUS, BERR, RSB, the Association of British Healthcare Industries, the Medical Research Council, the Engineering & Physical Sciences Research Council and the National Innovation Centre, working with patient groups and clinicians.

Other Initiatives

Pharmaceutical Industry

1. The then PM recognised the importance of the pharmaceutical industry to the UK when he announced the Pharmaceutical Industry Competitiveness Task Force (PICTF) in 1999, and the need to have a good relationship between the Government and industry. This relationship continues today through the Ministerial Industry Strategy Group (MISG).
2. The PICTF process set a new direction of travel in relation to the relationship the Government had with the pharmaceutical industry as it provided a structured, action-oriented platform for effective dialogue. It was recommended in the PICTF report (2001) that this close working should continue through the MISG. MISG is a high-level group and its role is to take a strategic overview of the UK environment. In order to reflect the structure of PICTF it is co-chaired by the relevant DH Minister and a company Chief Executive.
3. In 2005, MISG established a Long Term Leadership Strategy (LTLS)“to develop a long term strategy for medicines designed to: secure the provision of safe and effective medicines for patients; maintain and strengthen the UK pharmaceutical industry within Europe; and to advance healthcare innovation in the UK.”
4. The government and industry have been working together to take this Strategy forward. The Strategy has three main workstreams -which are looking at regulatory, European and partnership issues:
 - **Partnership Working Group** – partnerships between the NHS and pharmaceutical industry. It concentrates on uptake of medicines.
 - **European Working Group** - provides a mechanism for the Government and industry to develop proposals to assist the aims of the European Commission’s Pharmaceutical Forum to improve European competitiveness, and will in future deal with relevant wider international issues.
 - **Regulatory Working Group** - explores how to achieve and sustain a UK & European regulatory environment that will support the innovative European pharmaceutical industry and meet the needs of government, patients and prescribers.

The outcomes of the above conjoint efforts were published early on 6 February 2007, and work continues to implement the recommendations.

Healthcare Industries Task Force

5. The Healthcare Industries Task Force (HITF) was established in 2003 for a period of one year to explore areas where closer working between Government and the industry would result in benefits for all stakeholders. Led jointly by Lord Warner and Sir Chris O'Donnell, the Task Force included representation from across Government at Ministerial and senior level, and leading executives from the industry. As the first high level initiative between Government and this sector, it broke new ground and agreed nine key outputs, designed to improve the business environment for the industry whilst also improving patients' access to beneficial new medical technologies. The main objective was how to accelerate the adoption of beneficial new medical technologies and measures were devised to improve the NHS procurement process, the NHS environment for R&D on medical devices, workforce training and education, better support for medical technology innovators and the establishment of a technology adoption hub.
6. The HITF Strategic Implementation Group (SIG) was subsequently formed to oversee the start of the delivery of HITF outputs, and to continue to develop the dialogue between Government and the industry. A key issue continued to be how to balance the NHS's need to secure good value for money with investment in innovation where the returns are likely to be realised only in the longer term. Joint working therefore continued to develop a more systematic approach to the adoption process, including procurement, involving key organisations on the "innovation landscape".

Ministerial Medical Technology Strategy Group

7. In March 2006, Lord Hunt and Sir Chris O'Donnell published the SIG report outlining progress on HITF implementation, in which both Government and industry also committed to elevate their engagement to a strategic level. As a result, the Ministerial Medical Technology Strategy Group (MMTSG) was established in autumn 2007 to develop this agenda. MMTSG has shifted its focus towards developing a shared vision of how medical technology could achieve its potential for transforming health and social care services whilst also delivering good value. A key component of discussions is concerned with speeding the adoption process and facilitating more widespread diffusion. MMTSG is therefore contributing to the medical devices workstream of the Health Innovation Council (HIC).

**Progress on implementation of commitments in
Genetics White Paper (2003) and SofS' 2001 speech on genetics**

NHS genetics team, Department of Health February 2008

The Genetics White Paper, "Our inheritance, our future - realising the potential of genetics in the NHS", was published in June 2003. It set out a wide-ranging programme to help the NHS make appropriate use of genetic knowledge and technology as it becomes available, and announced £50m to fund this programme. These built on earlier commitments made by the then Secretary of State Alan Milburn in his speech on genetics in 2001, which included establishment of two National Genetics Reference Laboratories (see www.ngrl.org.uk) and the UK Genetic Testing Network (see www.geneticstestingnetwork.org.uk).

The following table sets out progress against each commitment.

Commitment	Progress to date
Chapter 2: Strengthening specialised services	
Laboratories:	
Invest up to £18 million capital 2003-2006 on upgrading NHS genetics laboratory facilities in England, in return for innovative plans for modernisation developed to meet local needs. As a result by 2006 genetic testing times will be cut and results should be available to following standards:	Bidding process completed and funds allocated during 2004-5 and 2005-6. New equipment installed including fast throughput and robotics to support quicker and higher volume testing.
<ul style="list-style-type: none"> • within three days where the result is needed urgently (e.g. for prenatal 	Significant progress in reducing waiting times while dealing with

<p>diagnosis)</p> <ul style="list-style-type: none"> • within two weeks where the potential genetic mutation is already known (e.g. because another family member has already been tested) • within eight weeks for unknown mutations in a large gene. 	<p>increased volumes of testing including the impact of the NICE clinical guideline on management of familial breast cancer.</p>
<p>Invest up to £1 million over three years in IT for genetics laboratories in the Genetic Testing Network</p>	<p>Output based specification and consultancy report on options made available to NHS stakeholders. £2 million funds allocated to NHS November 2006.</p>
<p>Workforce:</p>	
<p>Fund new clinical training posts to train over 50 new genetics counsellors over five years</p>	<p>First tranche have completed training; second tranche in training. Decisions currently being made about funding for final tranche.</p>
<p>Fund up to 90 new Grade A trainees in laboratory genetics and the equivalent of ten full-time trainer posts</p>	<p>Funding has been provided for four year trainee posts. Phased intake to match capacity of service to support training and take-on new staff. First intake now coming to end and fourth just starting, taking total trainee numbers up to 73. Trainer posts have been extended by 3 years to take them through to late 2010. We are now exploring with the Chief Scientific Officer how we can support development work to take forward modernising health care scientist careers in the genetics disciplines.</p>
<p>Chapter 3: Building genetics into mainstream services</p>	
<p>Pilot projects:</p>	
<p>Co-fund with Macmillan Cancer Relief pilots in up to six cancer network areas of model of delivering services for people at risk of, or concerned about, familial cancer</p>	<p>7 pilots, 3 of which have a primary care focus, have completed their final reports. Their experiences will be used as a basis for commissioning guidance to be issued as part of the implementation of the Cancer Reform Strategy 2007. Details of the lessons and experiences of these pilots are reported in a special edition of 'Familial Cancer' <i>Delivering Cancer Genetics Services – New</i></p>

	<i>Ways of Working 2007 vol.6(2)</i>
Pilot a systematic programme to identify and treat people with familial hypercholesterolaemia (FH)	This project has been managed by London IDEAS Genetics Knowledge Park. 11 lipid clinics in 5 sites were included to cover a range of catchment populations and organisational arrangements. The scheme piloted cascade testing for FH (tracing and testing of family members of known cases). A report of the results of the pilots and recommendations to the DH were published in July 2007 (available at www.fhcascade.org.uk/) Lessons from this work will help inform implementation of the planned NICE clinical guideline on the identification and management of familial hypercholesterolaemia.
Provide up to £2 million of start-up funding over three years for other initiatives to bring benefits of genetics into mainstream clinical areas; ditto specifically for primary care genetics initiatives	10 pilots have been completed, most of which set up innovative services to incorporate genetics advice and testing into mainstream medicine and to promote patient pathways integrating genetic advice with diagnosis treatment for patients suffering from familial disease. 3 of the services are continuing with funding provided by local commissioners pending further assessment of their effectiveness.
Provide start-up funding to allow up to ten GPs with a special interest in genetics to be established over the next three years	Framework document agreed with Royal Colleges; appointments made to 10 GP posts.
Evaluation of above pilot initiatives	Team at Nottingham University are undertaking an external evaluation (to complement internal evaluation by each pilot). They have published papers based on their interim findings. The final report covering generic organisational issues and challenges is expected Autumn 2008.
Screening:	
Two programmes, expected to be fully implemented in England by 2004/5,	

<p>will ensure that:</p> <ul style="list-style-type: none"> • all pregnant women are offered antenatal screening for Down's syndrome and are then counselled by midwives to help them make an informed choice • all babies are tested for hearing defects using the otoacoustic emissions test and/or auditory brain stem response 	<p>As part of the Fetal Anomaly Screening Programme, Down's syndrome screening is offered in almost all maternity units to women of all ages.</p> <p>The roll out of newborn screening is now complete and offered to all babies and 99% of newborn babies are screened.</p>
<p>In England by the end of 2004:</p> <ul style="list-style-type: none"> • A newborn screening programme will be in place offering screening for sickle cell disease • An antenatal screening programme for sickle cell and thalassaemia will be in place aiming to offer screening to around 200,000 pregnant women a year, initially targeting areas of high prevalence for these diseases. 	<p>Newborn screening for sickle cell is now fully implemented and cover the whole of England and is already identifying approximately 300 babies a year who would be at risk of death unless treated promptly.</p> <p>Antenatal screening for sickle cell and thalassaemia is expected to be rolled out in all trusts by Spring 2008.</p>
<p>Ask Human Genetics Commission to work with National Screening Committee to consider case for screening babies at birth</p>	<p>The report of the Joint Working Group of the Human Genetics Commission and the National Screening Committee, <i>Profiling the newborn: a prospective gene technology?</i> was published in March 2005. See www.hgc.gov.uk It concluded that genetic profiling of newborns was unlikely to be a practical or acceptable practice for the foreseeable future.</p>
<p>Chapter 4: Spreading knowledge across the NHS</p>	
<p>Set up an National Genetics Education and Development Centre (NGEDC)</p>	<p>The Centre (based at Birmingham Women's Hospital) started work in September 2004. See http://www.geneticseducation.nhs.uk</p>
<p>Set up Genetics Visiting Fellowships Fund</p>	<p>Four outgoing and two incoming fellowships have now completed their period of study and enable healthcare professionals in the NHS to learn from practice and experience in genetics around the world.</p>

<p>Develop genetics portal on the National electronic Library for Health....fund production of specially written material</p>	<p>Contract for dedicated team started July 2003; system live; renewal contract awarded in October 2007 to the National Genetics Education and Development Centre (NGEDC). Access the Genetics Conditions specialist library of the National Library for Health at www.library.nhs.uk/genepool/</p>
<p>Ensure NHS Direct keeps abreast of developments in genetics</p>	<p>There has not been a need for any specific work here to date.</p>
<p>Support evidence based care. The National Horizon Scanning Centre will be including genetics in its regular programme of work. The National Genetics Reference Laboratories will undertake health technology assessments. Some developments in genetics may also be included in the National Health Technology Assessment programme. DH will feed new developments in genetics, including pharmacogenetics, into prioritisation process for NICE's work programme as evidence base develops</p>	<p>Ongoing. Several genetics-related HTAs and evaluations by the NGRLs have been published. See the NGRLs' websites:</p> <p>NGRL(Manchester) www.ngrl.org.uk/Manchester/Technologypubs.htm NGRL(Wessex) www.ngrl.org.uk/Wessex/technologies.htm</p> <p>Since the White Paper's publication, NICE have published a guideline on familial breast cancer and have guidelines on other clinical areas of direct relevance to genetics in their work programme eg clinical guideline on the identification and management of familial hypercholesterolaemia.</p>
<p>Ensure that genetics is included in developments in NHS informatics such as the Integrated Care Records Service, minimum data sets, and clinical terms wherever this would be feasible and appropriate</p>	<p>Ongoing; work on clinical terms, data sets and messaging standards underway.</p>

Invest in training for commissioners	Successful events run in the South West and in London.
Chapter 5: Generating new knowledge and applications	
More details about the scope of research under the genetics White Paper and the individual projects that have been funded can be found on the DH Genetics Research Programme website http://www.genres.org.uk/index.htm	
Gene therapy:	
Invest up to £3 million to support gene therapy research on single gene disorders	3 projects selected and underway. Of these, 2 clinical trials are open to recruitment.
Fund research into the long-term safety of the use of gene therapy vectors which are designed to insert into human genetic material	4 projects selected and underway 3 of which are completed.
Provide £2.5 million over 5 years to support gene therapy research for cystic fibrosis	2 projects completed, a third project is underway with a clinical trial expected to open in 2008.
Invest up to £4 million to provide access to high standard facilities for gene therapy vector production NHS and other public sector researchers	Information-gathering exercise and expert workshop completed; three cancer trials supported by end of 2007, another 2 trials chosen for support (expected to begin in 2008); and further support is expected in 2008.
Pharmacogenetics:	
Invest up to £4 million to fund pharmacogenetic research on existing medicines	6 projects selected and underway.
Provide funding to set up first university Chair in pharmacogenetics, supported by a small department of two to three full-time researchers	Funding awarded to University of Liverpool for 5 years commencing September 2007. Professor Munir Pirmohamed appointed first NHS Chair in Pharmacogenetics.

Health services research:	
Invest up to £1.5 million to fund a range of research projects in area of genetics based health services	Following a priority-setting process involving stakeholders, 7 projects funded & underway.
When technology becomes available, provide up to £500,000 to support piloting of near patient genetic testing in NHS	Awaiting further progress with technology and clinical applications.
Publish an independent study into impact and management of intellectual property rights within healthcare sector	Report published July 2003, see http://www.phgu.org.uk/pages/work/IP.htm
Continue to monitor the conduct of genetic research in UK and take action to introduce checks and balances where necessary	Ongoing
Chapter 6: Ensuring public confidence	
...negotiated a moratorium with the insurance industry that will last until 2006.... committed to working with patient groups and with the industry to ensure a longer-term solution.	Published (in March 2005) Concordat and Moratorium on Genetics and Insurance, which extends the current moratorium until November 2011. Insurance industry have agreed not to submit any applications until 2008.
Continue to promote public understanding of genetics, including through additional financial support to Progress Educational Trust	Progress Educational Trust work programme now completed.
Provide around £1,000,000 to support a programme of events and initiatives in 2003 to celebrate 50th anniversary of discovery of structure of DNA.	DH contribution to the £1m was made available during 2003 to support a successful series of events and publications to celebrate the anniversary.
Ensure that regulatory framework anticipates and continues to address public concerns	Ongoing
Introduce legislation making it an offence to test person's DNA without	Under the Human Tissue Act 2005, it will be an offence to have

<p>their knowledge or consent except as part of medical treatment where consent is impossible to obtain, or use by police or courts</p>	<p>DNA for the purposes of analysing DNA without consent. The Act will come into force fully in September 2006.</p>
<p>Consider evidence for unfair discrimination against people on basis of their genetic characteristics and appropriate means of addressing any concerns in this area</p>	<p>Lord Sainsbury asked HGC to consider this issue with specific regard to employment aspects in 2005. These issues, along with others, are being considered in the context of the Government's broader review of discrimination legislation.</p> <p>Both HGC & GAIC continue to monitor genetic discrimination with regard to insurance. More information on the HGC's work on discrimination can be found at http://www.hgc.gov.uk/Client/Content.asp?ContentId=254</p>

Bodies contributing to the advisory framework for policy development

A number of these professional bodies or advisory committees will provide independent evidence to the Select Committee. In addition to those groups referred to below a number of professional and independent bodies, such as the Royal College of Physician's Committee on Ethics in Medicine, the British Medical Association's Ethics Committee, the Nuffield Council on Bioethics and the Public Health Genetics Foundation also make valuable contributions to informing the debate on the ethical, legal and social implications of genomic medicine.

Advisory Group on Genetics Research (AGGR) established by DH to provide expert advice on aspects of genetics research funded through the genetics White Paper. The responsibilities of the AGGR included: providing a strategic oversight of the programmes of genetics research funded by the DH and advising the DH Portfolio Director of Genetics Research on areas where additional research may be required to address the needs of the NHS and wider DH. These projects are now coming to an end and the group was wound up in 2007.

British Society for Human Genetics represents UK human genetics professionals. Its constituent organisations are: Clinical Genetics Society (CGS), Association for Clinical Cytogenetics (ACC), Clinical Molecular Genetics Society (CMGS), Association of Genetic Nurses and Counsellors (AGNC) and Cancer Genetics Group (CGG). www.bshg.org.uk

Chief Scientific Advisers Committee (CSAC) the principal committee at official level dealing with issues relating to science, engineering and technology (SE&T). CSAC considers issues of relevance to Her Majesty's Government and the devolved administrations concerning SE&T.

Council on Science and Technology (CST) is the UK Government's top-level advisory body on strategic science and technology policy issues. CST's remit is to provide independent advice to the Prime Minister, the First Ministers of Scotland, Wales and Northern Ireland on strategic issues that cut across the responsibilities of individual government departments.

Genetics Commissioning Advisory Group (GenCAG) set up 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. Members are drawn from relevant professional bodies and groups and Royal Colleges, an umbrella patient support group (the Genetic Interest Group - GIG) and from the specialised commissioning groups which commission these services in the NHS. www.dh.gov.uk/Genetics/

Gene Therapy Advisory Committee (GTAC) is the UK national research ethics committee for gene therapy research under the Clinical Trials Regulations and a ministerial advisory body for gene therapy research and its implications. The UK is the only European country that has a dedicated body to give advice on and provide ethical oversight of gene therapy clinical research. www.advisorybodies.doh.gov.uk/genetics/gtac/index.htm

GTAC's work has helped foster UK gene therapy, making the UK lead gene therapy research in Europe with over 40% of trials, second only to the US worldwide²⁹. With support from the Genetics White Paper, two novel gene therapy trials, the first of their kind in the UK, are now underway. Patients with two debilitating single gene disorders, Duchenne Muscular Dystrophy and a form of childhood blindness are being recruited as well as a new trial for Cystic Fibrosis, the most common single gene disorder in the UK.

Genetics and Insurance Committee (GAIC) considers applications by the insurance industry for the use of predictive genetic test results in setting insurance premiums, and provides advice to Health, Science and Treasury Ministers. GAIC also has a number of supervisory functions of the insurance industry under the 2005 Concordat and Moratorium on Genetics and Insurance³⁰. www.advisorybodies.doh.gov.uk/genetics/gaic/index.htm

Human Fertilisation and Embryology Authority (HFEA) All clinics and research centres in the UK, whether private or NHS based, wishing to undertake treatments, services or embryo research covered by the HFE Act must hold a licence from the HFEA. The range of fertility treatments and services regulated by the HFEA was extended on 5th July 2007, when the Human Fertilisation & Embryology (Quality & Safety) Regulations 2007 amended the 1990 Act to bring the EU Tissues and Cells Directive into force in respect of reproductive cells. From that date, establishments carrying out the donation, procurement, testing, processing, preservation and distribution of reproductive cells also require a licence from the HFEA. www.hfea.gov.uk

Human Genetics Commission (HGC) UK Government's advisory body on new developments in human genetics and how they impact on individual lives. It advises the Government on human genetics with a particular focus on the social, ethical and legal issues associated with new scientific developments and genetics-related innovations in medicine and other fields such as forensics, insurance and employment. It has a broad membership including bioethicists, lawyers and social scientists. The HGC reports to Health and Science Ministers and Ministers in Scotland, Wales and Northern Ireland. Its terms of reference include a role in advising on strategic priorities for research and the delivery of genetics services by the NHS. www.hgc.gov.uk

²⁹ Edelstein *et al*, (2007) *J Gene Med*, Oct;9(10):833-42

³⁰

http://www.dh.gov.uk/en/Publicationsandstatistics/Publications/PublicationsPolicyAndGuidance/DH_4105905

National Genetics Reference Laboratories (NGRLs), were first established in 2002 to support the network of NHS genetics laboratories. Their key roles are to:

- Promote and help take forward the development of genetic laboratory science and practice in the NHS.
- Act as a dedicated resource to support the NHS molecular and cytogenetic communities in their development of better patient services.

They have a particular role in evaluating new technologies and developing new techniques and in leading work to develop informatics and appropriate quality assurance (QA) systems. They are sited in Manchester and Wessex (Salisbury). Following a successful review of their functions their contracts have been renewed for a further five years taking them through to 2012. www.ngrl.org.uk

NGRL Steering Group has been established to oversee the work of the NGRLs during the new contractual period. It is chaired by Professor Martin Bobrow and draws its membership from a wide range of professional bodies and organisations across many laboratory disciplines. As well as advising DH on the work programme and priorities of the NGRLs it advises DH more broadly on developing technologies that will impact on NHS laboratory services in the future. Its key role is:

- To take a strategic view of developments and innovations which may be relevant to NHS genetic laboratory service
- To advise the DH on the implications of such developments for the strategic direction of the National Genetics Reference Laboratories (NGRLs) and of the NHS genetic laboratory service more generally.

Joint Committee on Medical Genetics (JCMG) made up of the Royal Colleges of Physicians, Pathologists, of Obstetricians and Gynaecologists and of General Practitioners and the BSHG.. Its remit includes “discussing and co-ordinating advice to Government and other bodies on policy and services issues relating to genetics in medicine.” DH has observer status and officials regularly attend JCMG’s meetings.

www.bshg.org.uk/joint_committee/joint_committe.htm

UK Genetic Testing Network (UKGTN) collaborative group of laboratories and their clinicians, commissioners and patient representatives involved in the assessment of provision of genetic tests for inherited disorders. It advises policy development and the NHS through the UK GTN Steering Group and a number of working groups. www.ukgtn.nhs.uk

Horizon Scanning

1. Effective horizon scanning allows the Government departments to ensure that the scientific cutting edge in any given area can lead to improved interventions for health and social care. It also helps to ensure the Government departments are thinking about the future when developing policy and is commissioning the right science to meet the future needs of the population.

DH Horizon Scanning

DH Horizon Scanning Unit

2. The Department's specialist Horizon Scanning Unit was launched in July 2007, in response to a decision by the Policy Committee to increase the emphasis in policy development on future-proofing policies and ensuring robustness. The HSU's main aim is to improve horizon scanning capability within the Department and to use this to improve the work of the Department. As such, the idea that horizon scanning should always be relevant to the work of the Department and useful for policy-making is integral to the work of the Horizon Scanning Unit. Indeed, one of the Unit's objectives states explicitly that it should,

“Ensure horizon scanning and futures work is useful to the Department and improves its work, including its resilience to risk and its ability to take advantage of untapped opportunities for innovation.”

3. As part of the development of the Horizon Scanning Unit independent horizon scanning functions, such as those described in this section, were brought together into a Horizon Scanning Network. This network involves collaboration of officials and experts from across the Department, and liaises with horizon scanning bodies in other Government Departments, such as Foresight. In addition, the Horizon Scanning Unit has set up a site on the Department's intranet (Delphi) as a corporate resource for sharing scanning information, contacts and methodology.

4. The Horizon Scanning Unit works with policy teams in three broad ways:

	1	2	3
How does the Unit work with other teams?	The HSU coordinates work, receiving input from teams within Directorates and external futures networks	The HSU works with the teams embedded within Directorates on specific, commissioned topics	Horizon Scanning teams embedded within Directorates work with support from the HSU

On what processes?	1. Scanning the horizon for early signals 2. Tracking trends and driving forces	Scoping futures	1. Testing policies for resilience to risk 2. Exploring untapped opportunities for innovation
To produce what products?	Chart-books of key trends; horizon scanning section on Delphi (DH intranet)	Scenarios and vignettes of possible, plausible and preferred futures	Reports and presentations to Top Of The Office (TOTO) and the Senior Leadership Team (SLT) of key findings

5. The development of the Unit helps demonstrate the importance the Department attaches to horizon scanning and the pervasive role we believe it should play in our work. We are committed to effective horizon scanning as a vital strand of good policy-making. The Horizon Scanning Unit has produced an initial work programme and has already produced its first report.

Horizon scanning in subject areas

6. Even before the establishment of the Horizon Scanning Unit, horizon scanning was closely tied to the development of strategy and policy. The Strategy Unit's report into Long-Term Conditions was a vital piece of horizon scanning to help the Department understand the problems it would face over the upcoming years and decades. The report identified patterns of growth in the number of patients with long-term conditions and projected the future of this growth, further identifying factors that could affect this projection, such as a rise in obesity and diabetes and an ageing population. This kind of fundamental horizon scanning is important and valuable not only for the report for which it was conducted but also because it can provide evidence about the future state of the population, on which other policy teams can build.³¹

7. Horizon scanning of this broader nature can provide a context in which topic-specific horizon scanning can be better understood. The Department undertakes this kind of specialised horizon scanning in a number of ways – whether through sponsorship or direct action – but always with the horizon scanning being conducted by those with the most appropriate expertise. For example, the National Horizon Scanning Centre (NHSC) at the University of Birmingham scans the horizon for new and emerging health technologies. The NHSC's findings are fed into the early part of the topic selection process for NICE's consideration panels to assess. Potential topics for NICE's work

³¹ *Accelerating Progress in Long-Term Condition Management* (September 2007) Department of Health internal document – attached

programme are then recommended to the DH and Ministers to consider formal referral to NICE to develop guidance.

8. The National Horizon Scanning Centre Research Programme is part of the National Institute for Health Research (NIHR) and is funded by the Department. The Centre investigates new and emerging health technologies that may require urgent evaluation and provides advance notice to the Department. The Centre also has an international network of other horizon scanning organisations with which it works. It is a member of both the International Information Network for New and Changing Health Technologies (Euroscan) and the International Network of Agencies for Health Technology Assessment.
9. It is vitally important to the Department that horizon scanning within this field is translated into applied research that can have a tangible benefit for the NHS. The New and Emerging Applications of Technology Programme (NEAT) – part of NIHR – supports strategic and applied research into the use of new or emerging technologies, the outputs of which must be generalisable and capable of being applied to a defined health or social care need.³²
10. Horizon scanning for advances in scientific technology has also helped to place the UK at the forefront of the fast-developing arenas of stem-cell and genetic science. To respond to the rapidly changing environment, the Department needs to:
 - ensure an effective strategic advisory and regulatory structure that identifies and maximises benefits from potential advances in human genetics;
 - address broad ethical, legal and social implications arising from advances; and
 - manage the process of change as practical applications of advances are introduced, especially in the NHS.
11. To achieve this, the Government needs advice from a variety of sources. These sources will approach horizon scanning from their own, particular area of expertise but, partly through the Horizon Scanning Unit, these different elements will complement each other to provide a fuller picture.
12. Through the establishment of the Human Genetics Commission (HGC) the Department has ensured that Ministers and officials receive expert advice on current and potential developments in human genetics, their likely impact on human health and healthcare and their social, ethical, legal and economic implications. The HGC also advise on strategic priorities for research and in the delivery of genetic services by the NHS. The HGC is committed to debating these issues in an open and transparent way and holds open meetings and publishes its advice. The HGC's working groups examine

³² Further information on NEAT is available at www.nihr-ccf.org.uk/site/programmes/neat/

issues at the very forefront of genetic science – issues such as reproductive decision-making, on which they published their report *Making Babies*.³³

13. The Department has been involved in horizon scanning in this area for some time. In 2000, the Department published a report entitled *Stem cell research: Medical progress with responsibility*. This report was produced by an Expert Group established by DH and chaired by the Chief Medical Officer. The Group was asked to undertake an assessment of the anticipated benefits of new areas of research using human embryos, the risks and the alternatives and, in the light of that assessment, to advise whether these new areas of research should be permitted.

14. In addition, the Group analysed the Human Fertilisation and Embryology Act (1990) and found that it still provided “the necessary safeguards against the inappropriate use of embryos in research.” The report also found that, “the manner of regulating any proposed research within the UK is sufficiently finely tuned to be able to take account of particular ethical concerns. Indeed, the UK enjoys a leading international position in the resolution of these difficult questions.”³⁴

15. More recently, the establishment of the UK Stem Cell Initiative (UKSCI) was announced in March 2005. The initiative, which reported jointly to DH, DIUS and HMT, was charged with (amongst other things) developing a ten-year vision for UK stem cell research, to make the UK the most scientifically and commercially productive location for this activity over the coming decade. UKSCI published its report in 2005 – the Pattison report. The report “acknowledged the appropriateness of the UK system of regulatory oversight of embryo research and the investment that has been made to date across research funding, infrastructure, people and support for private investment.”³⁵ The Government accepted the report’s recommendations and as a result an additional investment of £50 million was provided over the period of 2006-07 and 2007-08 by the public sector.

16. The 1990 Act is currently being revised, in part to take account of new scientific and clinical developments. The Department relies heavily on horizon-scanning functions embedded within the Human Fertilisation and Embryology Authority, as well as on advice from expert groups of the Medical Research Council and the Academy of Medical Sciences. Examples include detailed analyses of technologies for sex selection by sorting of human gametes and the creation of human-animal hybrids.

17. As part of the response to the Pattison report, the Government asked the Gene Therapy Advisory Committee (GTAC) in 2006 to conduct the ethics

³³ *Making Babies: reproductive decisions and genetic technologies* (January 2006) Human Genetics Commission www.hgc.gov.uk/UploadDocs/DocPub/Document/Making%20Babies%20Report%20-%20final%20pdf.pdf

³⁴ *Stem Cell Research: Medical progress with responsibility* (June 2000) Dept of Health <http://www.hgc.gov.uk/UploadDocs/DocPub/Document/Making%20Babies%20Report%20-%20final%20pdf.pdf> – attached

³⁵ Government response to the UK Stem Cell Initiative report and recommendations (2006) http://www.dh.gov.uk/en/Policyandguidance/Healthandsocialcaretopics/Stemcell/Stemcellgeneralinformation/DH_4124082

review of certain stem cell trials. This oversight role was to build on GTAC's specific expertise as the UK national ethics committee for gene therapy research, and adds to GTAC's existing roles as a Ministerial advisory body, and an outward-facing advisory body. The Committee also provides a horizon scanning function, which feeds into a wider forum than advice to the Department. GTAC's New and Emerging Technologies (NET) subgroup has a horizon-scanning function specific to technological developments in the area of gene therapy and related technologies.

18. An example of how GTAC's horizon scanning activities have led to practical guidance that has been accepted internationally was GTAC's report on the potential use of gene therapy *in utero*. GTAC's NET subgroup considered *in utero* gene therapy, its possible consequences and implications. Following the NET subgroup's report, GTAC concluded publicly "that the use of a direct, or vector, mediated gene therapy *in utero* are unlikely to be acceptable for the foreseeable future, in view of the safety and ethical difficulties."³⁶ This advice still holds true and no *in utero* procedures have been performed in the UK or elsewhere in the world.

19. More recently, GTAC's horizon scanning activities have brought to light "the development of liver tumours in a pre-clinical study using lentiviral vectors." In response to this, GTAC issued an open letter on its views, published on its website.³⁷

20. Another area in which the Department benefits from scientific expertise specifically focused on horizon scanning is infectious diseases. The National Expert Panel on New and Emerging Infections (NEPNEI) was set up by the Department in 2003, having been proposed in the Chief Medical Officer's (CMO) Infectious Disease Strategy, *Getting Ahead of the Curve*.³⁸ NEPNEI's principal role is to identify emerging and potential infectious threats to public health both nationally and internationally and provide advice to CMO. The secretariat is provided by the Department and the Health Protection Agency (HPA). The Panel's Terms of Reference are to identify key areas for action and to advise on priorities by:

- Identifying emerging and potential infectious threats to public health both nationally and internationally;
- Placing emerging infections in the wider clinical and public health contexts;
- Advising on prevention and control measures;
- Prioritising areas for surveillance and for information needs;
- Advising on research, including technological development needs.

³⁶ Report on the potential use of gene therapy *in utero*, November 1998, GTAC <http://www.advisorybodies.doh.gov.uk/genetics/gtac/inutero.htm> - attached

³⁷ Open letter entitled 'The UK Gene Therapy Advisory Committee (GTAC) has concerns regarding the safety of some lentiviral gene therapy vectors', 5th November 2004 <http://www.advisorybodies.doh.gov.uk/genetics/gtac/lentiviruses-1104.pdf> - attached

³⁸ *Getting Ahead of the Curve: A strategy for infectious diseases* (2003) Dept of Health http://www.dh.gov.uk/en/Consultations/Closedconsultations/DH_4016942

21. Examples of areas where NEPNEI has been involved include the development of the UK strategy for dealing with pandemic influenza; the establishment of a clear mechanism for commissioning and funding of research to ensure that research is not delayed in the event of an outbreak of a new emerging infection; and the prioritisation of actions required to tackle the growing problem of antimicrobial resistance.
22. Whilst the paragraphs above provide only a snapshot of the horizon scanning work undertaken by the Department – and a snapshot from only a few, specific areas – they demonstrate the Department’s understanding that embedded horizon scanning, when underpinned by a coherent network such as that provided by the HSU, results in relevant, cutting-edge horizon scanning that can be used to develop policy. However, we are not complacent. Having established the Horizon Scanning Unit, we are now looking at some practical proposals to improve capability and engagement across the Department, and these ideas were considered at the most recent Policy Committee meeting.

DIUS Horizon Scanning

23. One of the societal or economic Grand Challenges that the Research Councils announced in December 2007 will address lifelong health and well-being, in response to the the Treasury’s Grand Challenge on the effects of an ageing population on economic competitiveness. The Research Councils’ Grand Challenges will be developed in conjunction with researchers, users and the public and will include basic research through to application plus studies on risk governance, economics, and social implications.
24. The Horizon Scanning Centre³⁹ in the Government Office for Science has been running a Wider Implications of Science and Technology⁴⁰ (WIST) programme of stakeholder engagement to identify the safety, health, environmental, ethical, regulatory and social implications of new and emerging areas of science and technology. This has been closely integrated with a Department of Innovation, Universities and Skills funded Sciencewise⁴¹ programme (sciencehorizons⁴²) of public engagement on emerging technologies. Both the stakeholder and public engagement programmes covered all areas of emerging science and technology so the findings are correspondingly wide-ranging. They include the expectation that anticipated developments in the next 15 – 20 years will enable a patient to know or have the opportunity to be appraised of important health-related

³⁹ http://www.foresight.gov.uk/HORIZON_SCANNING_CENTRE/index.html

⁴⁰ http://www.foresight.gov.uk/HORIZON_SCANNING_CENTRE/WIST/Index.html

⁴¹ <http://www.sciencewise.org.uk/>

⁴² http://www.sciencewise.org.uk/html/projects.php?source=projectdetail&project_ID=9

features of his or her genome. Potential implications highlighted by the programme are –

- changed demands on health professionals and resources, and personalised, sophisticated treatments for individuals, possibly leading to data protection and ethical issues;
- the availability of better, earlier, personalised health information outside traditional healthcare settings (eg in supermarkets, virtual worlds and other settings, both state and commercial) offering the opportunity for people to take steps to prevent ill-health;
- how to balance a child's right *not* to know about his or her likely future health trajectory against the potential for early intervention to divert or prevent the onset of disease; and
- an expanding cohort of people that are unable to obtain health insurance and thus a need for regulation to restrict the information to which companies are allowed access.

25. The Foresight Programme within the Government Office for Science uses robust evidence and rigorous futures methodologies to identify and understand the risks and opportunities that arise from uncertainty in order to improve strategy across Government. Foresight's major projects address selected topics where developments in science and technology either pose a challenge or offer opportunities to the UK. The Horizon Scanning Centre uses a more eclectic range of evidence to address the entire spectrum of public policy futures - not just science and technology – in smaller projects, and also works to raise futures capability across Government.

MHRA and the in vitro medical devices (IVD) directive

1. As far as the Medicines and Healthcare products Regulatory Agency (MHRA) is aware there is currently no genetic test placed on the market as a medicine. In the event that a manufacturer applied for a medicines marketing authorisation then they would need to meet the licensing requirements under the Medicines Act. In this context, there is therefore no regulatory gap for these types of products.
2. Firstly it should be noted that the Medical Devices Regulations only cover genetic test kits which are placed on the market with a medical purpose which is determined by the definition of an IVD in the Directive. The Regulations cover kits placed on the market to test blood and tissue donations for the purpose of providing information on pre disposition to certain conditions or diagnosis of certain medical conditions. The Regulations do not regulate those tests which are for a non medical purpose such as those for paternity testing or for making general lifestyle choices such as determining dietary or fitness regimes. In addition the Regulations have no competence in the sense of influencing or otherwise the development of new or emerging technologies as far as genetic test kits are concerned
3. As far as the Agency is concerned genetic tests which use genome variation data and proteomic marker based tests/assays are covered by the requirements of the Regulations provided that they are testing for a medical condition.
4. The underlying principle of the IVD Directive is to remove technical barriers to trade by providing manufacturers with a single set of regulatory requirements that, once met, would provide free and unhindered access to the EU market. At the same time the Directive aims at providing users and patients of IVD's with a high level of confidence that, when used in accordance with the manufacturer's instructions, they are safe or an acceptable quality and performs as specified by the manufacturer. In outline the Directive is intended to ensure that IVD's do not compromise the health and safety of patients, users, and third parties and attain the performance levels attributed to them by their manufacturer.
5. In general terms a manufacturer wishing to place his IVD on the EU market must assign his device to one of the relevant risk categories defined in the Directive, ensure that the device meets the relevant essential safety requirements specified in the Directive and follow the appropriate conformity assessment procedure. The In vitro Diagnostic Medical Devices Directive has four risk classifications associated with risk associated with relative dangers to public health and/or patient treatment by the kit not performing as intended. The highest risk includes test kits for HIV, HTLV and Hepatitis and some blood grouping products including those to test donated blood. The second group covers test kits for rubella, toxoplasmosis and phenylketonuria

as well as self test kits for blood glucose. The third Group cover self test kits whilst the final group covers all other general test kits. Notified Body intervention (which is a third party independent certification organisation) is required for all the higher risk tests and the self test element of the kits used by the public in the home environment. The Notified Body will issue an EC Certificate of Conformity on the basis of an appropriate conformity assessment procedure in the Directive. In the main genetic test kits will fall within the third general risk category together with tests such as cholesterol and pregnancy.

6. The Agency is not aware of any genetic kits being made available over the counter for use by the general public in the home environment. However if any became available they would have to be designed and manufactured in such a way that they perform appropriately for their intended purpose taking into account the skills and means available to users and the influence resulting from variation that can reasonably be anticipated in users technique and environment. The information and instructions provided by the manufacturer should also be easily understood and applied by the user tacking into account their level of knowledge. Furthermore for self test IVDs the manufacturer is required to obtain certification from an independent conformity assessment organisation known as a notified body that would assess the design of the device in terms of its suitability for non-professional users
7. The majority of genetic tests used are assays put together in the hospital laboratory and used in that healthcare establishment's premises on their own patients. This is considered to be in-house manufacturing and because there is no placing on the market is not covered by the provisions of the Directive. If an in-house laboratory tests samples received from another healthcare establishment using their own assays again this is not regarded as placing on the market. That said such establishments are encouraged to manufacturer their assays in accordance with the requirements in the Directive. What needs to be clear is that the Directive regulates the test kit not the service provider.
8. In addition there are a number of commercial genetic testing service providers who provide a range of services on the basis of a supplied sample providing data for linkeage analysis, carrier screening, prenatal diagnostic testing, newborn screening, forensic testing, confirmational diagnostic testing i.e. for leukaemia, etc. Whether these tests or assays are regulated under the terms of the IVD Directive is determined by whether the reason for generating the data comes within the definition of an IVD as indicated above. The underlying principle of the IVD Directive is to remove technical barriers to trade by providing manufacturers with a single set of regulatory requirements that, once met, would provide free and unhindered access to the EU market. At the same time the Directive aims at providing users and patients of IVD's with a high level of confidence that ,when used in accordance with the manufacturer's instructions, they are safe or an acceptable quality and performs as specified by the manufacturer. In outline the Directive is intended to ensure that IVD's do not compromise the health

and safety of patients, users, and third parties and attain the performance levels attributed to them by their manufacturer.