

Our Inheritance, Our Future

**Realising the potential of genetics
in the NHS**

Presented to Parliament by the Secretary
of State for Health

By Command of Her Majesty

June 2003

Any comments or queries on this document should be addressed to Genetics White Paper, Genetics Branch, Department of Health, Room 651C, Skipton House, 80 London Road, London SE1 6LH. Email: Geneticswhitepaper@doh.gsi.gov.uk



DEPARTMENT OF HEALTH

Our Inheritance, Our Future

Realising the potential of genetics in the NHS

*Presented to Parliament by the Secretary of State for Health
By Command of Her Majesty
June 2003*

© Crown Copyright 2003

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

Any enquiries relating to the copyright in this document should be addressed to The Licensing Division, HMSO, St Clements House, 2-16 Colegate, Norwich, NR3 1BQ.
Fax: 01603 723000 or e-mail: licensing@cabinet-office.x.gsi.gov.uk

Foreword by the Prime Minister



Our country has a remarkable scientific tradition. The extraordinary achievements of Newton, Darwin and a host of other eminent scientists have both greatly increased the understanding of our world and improved the quality of life for everyone.

Our record continues to be outstanding; with just one per cent of the world's population, we receive nine per cent of scientific citations. Nowhere has this record been more notable in recent decades than in bio-science and bio-technology.

The discovery in Britain of the structure of DNA 50 years ago – perhaps the biggest single scientific advance of the last century – marked the beginning of a golden age of bio-science in Britain which continues today. It is likely to have as big an impact on our lives in the coming century as the computer had for the last generation.

The more we understand about the human genome, the greater will be the impact on our lives and on our healthcare. As an increasing number of diseases are linked to particular genes or gene sequences, we will be able to target and tailor treatment better to offset their impact and even to avoid the onset of ill-health many years in advance.

I am proud to know that much of this ground-breaking work is already taking place in our country. I am also absolutely determined that the National Health Service should be able to respond to these advances so the benefits of genetics and the more personalised and improved healthcare it will bring are available to all.

It means we must prepare now for the future. We must invest in research and research facilities to drive further discovery. We must ensure the NHS has the skills and expertise to make best use of these advances. And we must also be ready to explain clearly and consistently to patients the new health care choices this will bring. Through the independent experts on the Human Genetics Commission we will ensure that people have access to advice about human genetics and the social and ethical issues involved. This is the best way of easing understandable public anxiety about the coming revolution in health care.



This White Paper sets out how the Government intends to achieve all these goals. It also underlines our determination to ensure the NHS and its patients get the maximum benefit from the pioneering work already under way and the transformation it will bring in the delivery of health care in this country.

A handwritten signature in black ink that reads "Tony Blair". The signature is written in a cursive style and is positioned above a solid black horizontal line.

Tony Blair

Contents

Foreword	5
1. Recognising the potential	7
2. Strengthening specialised services	24
3. Building genetics into mainstream services	36
4. Spreading knowledge across the NHS	47
5. Generating new knowledge and applications	59
6. Ensuring public confidence	72
Annex A – Genetics services in Scotland, Wales and Northern Ireland	87
Annex B – Advisory Panel	89
Glossary	90



Foreword



Advances in human genetics will have a profound impact on healthcare. Over time we will see new ways of predicting and preventing ill health, more targeted and effective use of existing drugs and the development of new gene-based drugs and therapies that treat illness in novel ways. Above all, genetics holds out the promise of more personalised healthcare with prevention and treatment tailored according to a person's individual genetic profile.

The Government recognises and welcomes the potential offered by genetics to improve our health and healthcare. Britain's academic and industrial research prowess means that we are already amongst the leading players in genetics research and development. We are determined to harness that potential and to ensure that the benefits of genetics are realised throughout the NHS.

In April 2001, the Government announced new investment of £30 million to develop our specialised genetics services. This is already enabling them to see more patients and improve genetic testing provision. But there is much more to be done if the NHS is to be ready to cope with future demand and if genetic based healthcare is to expand and permeate every area of medicine as new genetics tests and therapies come on stream.

Our vision is 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. Over the next three years, the Government will invest an additional £50 million in England in developing genetics knowledge, skills and provision within the NHS. This document sets out how that money will be spent. By building on our strengths and starting to prepare now, genetic advances can deliver real and lasting benefits in health and healthcare for all of us.

A handwritten signature in black ink, which appears to read "John Reid". The signature is written in a cursive style and is positioned above a horizontal line.

Secretary of State for Health



This White Paper sets out a plan of action and investment in genetics for the NHS in England, as well as covering wider UK genetics policy issues. Activity on genetics health services in Scotland, Wales and Northern Ireland is summarised at Annex A.

1. Recognising the potential

- 1.1** Our genes play a fundamental role in determining our health and our response to healthcare. Six out of ten people are likely to develop a disease that is at least partially genetically determined by the age of 60. Greater knowledge of genetics will have a major impact on our understanding of human illnesses and herald a step-change in disease prevention, diagnosis and treatment. Although there are difficult moral issues raised by genetics advances we see enormous overall potential benefits for patients.
- 1.2** The year 2003 marks the 50th anniversary of Crick and Watson's discovery of the double helix structure of DNA – the “molecule of life” from which our genes are made. It also marks the year in which the Human Genome Project has been completed. The three billion chemical letters that make up DNA have been fully sequenced, and this will allow all the 30,000 plus genes in the human genome to be identified. The UK is a leading player in this ambitious international project with a third of the genome being mapped at the Wellcome Trust Sanger Institute in Cambridge.
- 1.3** The Human Genome Project will pave the way for a revolution in healthcare. Research is already underway to turn the knowledge gained into new treatments for cancer, heart disease, diabetes and other conditions. Over the next few decades we will learn more about:
- how individual genes, or groups of genes interacting together, can make us more predisposed to certain diseases. And how external factors such as smoking, diet or infection can interact with our genetic make-up to make the development of disease more likely
 - how some versions of genes can actually protect us from disease



- how our genes can affect our response to medicines. Pharmacogenetics will lead to prescribing which is more effectively tailored to the needs of the individual
- how genetic testing can become an integral part of health care. It will become faster, more accurate and cheaper. Testing in GP surgeries or local pharmacies will become a real possibility
- how diseases such as cancer work at a cellular level. This will lead to new treatments that target the disease rather than just control the symptoms, bringing new hope to many patients.

1.4 Here in Britain we start with a great advantage. We have in this country some of the best scientists, academics and universities. A great deal of the research and innovation into genetics is happening here – in both our pharmaceutical and biotechnology industries as well as in the public sector. Our NHS genetic services are admired throughout the world. We are well placed to lead the world in the discovery and realisation of the maximum benefits of genetics in healthcare.

1.5 The values of the NHS are particularly suited to capturing the benefits of genetic advances. The basis of the NHS – universal health care funded from taxation – protects people against discrimination on the basis of genetic information. As genetic testing becomes more widely available, people will not have to fear that they will lose cover for health care. Everyone regardless of their risk is “insured” by the NHS. Private health insurance usually bases how much you pay on how likely is the risk of your needing health care in future. Using genetic information private insurers, unless heavily regulated, could exclude benefits from policies, increase premiums (rendering them unaffordable for many) or simply refuse to insure people. In contrast, the values of Britain’s NHS mean citizens can choose to take genetic tests free from the fear that should they test positive they face an enormous bill for insurance or become priced out of cover altogether. It provides a bulwark against the inequalities and inefficiencies of private insurance systems where the prospect of a “genetic underclass” of the unwell and uninsurable, unable to pay the premiums for medical care, is a very real threat. Already in the United States of America, where 40 million people have no medical cover, developments in genetics have stirred precisely these concerns. As our understanding of genetics advances, the case for private health insurance as an alternative to our universal tax financed NHS diminishes.



Genes: what they are and how they work

Nearly all of our characteristics, from the colour of our eyes to our response to infections, are at least partly determined by our genes. Genes are the blueprint for making and running the human body. They carry information that tells cells how to make proteins – the basic building blocks of the human body. Almost all of our cells contain a complete set of the estimated 30,000 genes that make up the human genome.

Genes are made up of a substance called DNA. DNA looks like a long spiral staircase – the famous double helix. Each gene is a section of this DNA, like a piece of music on a cassette tape.

Within each cell, the long strands of DNA are tightly packaged into structures called chromosomes. Our cells have 23 pairs of chromosomes, one of each pair inherited from each of our parents when the 23 chromosomes in our mother's egg combined with the 23 chromosomes from our father's sperm. In this way we inherit one set of chromosomes – and hence one set of genes – from each parent.

Despite the fact that at the genetic level we are 99.9% the same, we are each of us genetically unique (except for identical twins). We owe our uniqueness to small differences in the remaining 0.1% of our genetic material. Some gene variations lead to only small differences between people, such as different blood group or height. Other differences are more important and may lead to disease.

(continued overleaf)



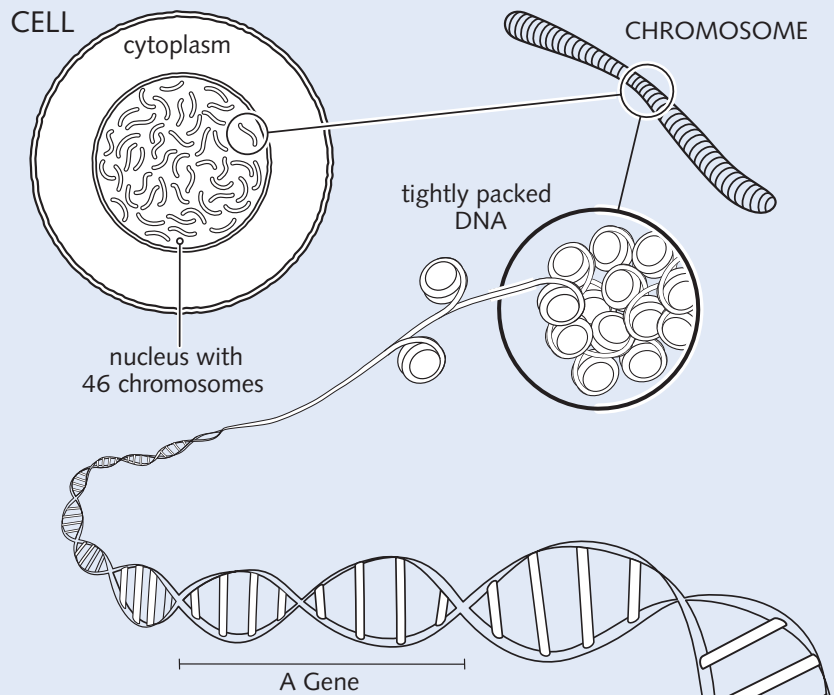
Genes: what they are and how they work – continued

Mutations are changes in the structure of DNA that can occur spontaneously during our lives or as a result of exposure to things in the environment such as toxins or radiation. Most of these are fixed by special repair machinery in the cell. If uncorrected, mutations can accumulate in a cell during a person's lifetime and they may lead to diseases such as cancer. If mutations affect sperm or egg cells or occur around the time of conception they may be passed down from parent to child. Not all mutations are bad. Some can actually protect us from disease.

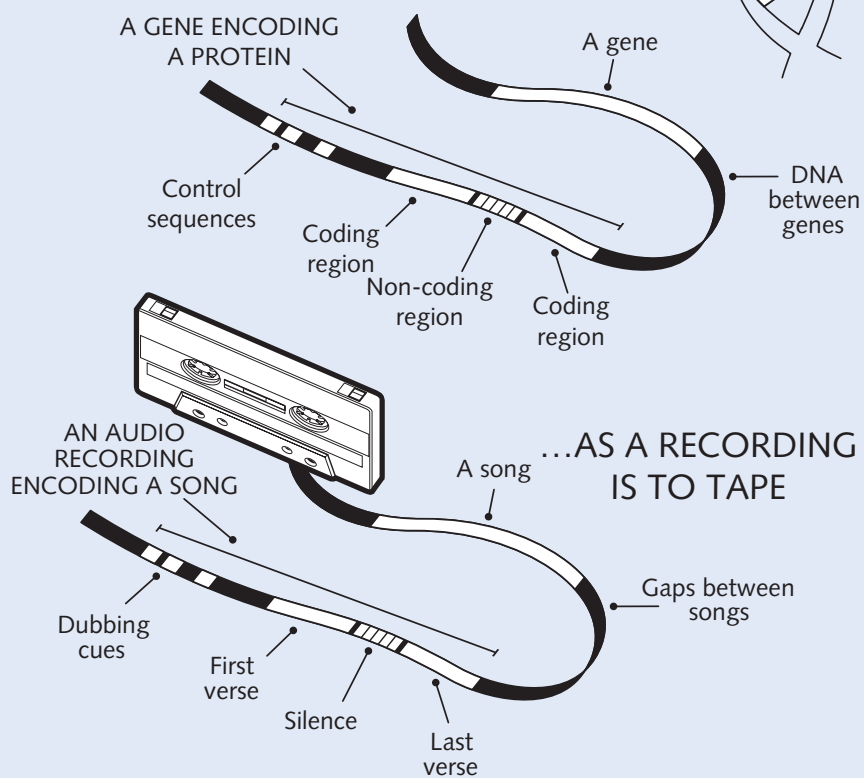
Some genetic mutations are known as “dominant”. In these cases, inheritance of the mutation from one parent only can be sufficient to cause the disorder. An example is Huntington's disease. Other mutations are “recessive” and the mutation has to be inherited from both parents for the person to be affected. Such parents are usually unaffected carriers because they only have a single copy of the gene – and may well not know that they have it. Common recessive disorders are cystic fibrosis and sickle cell disease.



A gene is a stretch of DNA like a piece of music on a tape



A GENE IS TO DNA...





Genetic testing

1.6 At present, most NHS genetics work is concerned with inherited disorders caused by a defect in a single gene or chromosome that results in a specific medical condition or syndrome. Examples of these disorders are cystic fibrosis, haemophilia and muscular dystrophy.

1.7 Genetic testing is currently used to:

- confirm a diagnosis where symptoms already exist
- indicate whether someone with a family history of a late-onset disease such as Huntington's is likely to develop the disease
- check whether someone is a carrier for a recessive disorder (where abnormal genes must be inherited from both parents to lead to the condition) such as cystic fibrosis
- screen before birth for genetic disorders such as Down's syndrome
- screen new born babies for genetic disease such as PKU (phenylketonuria).

1.8 There are around 10,000 single gene disorders. Most are rare, but taken together they are estimated to affect up to 5% of the population and cause considerable ill health and premature mortality. The treatment and care of patients with these disorders also costs health and social services about £2 billion each year.

1.9 Genetic tests now exist for about 200 of these disorders and more are being developed. The accuracy and potential impact of the diagnosis varies with the gene, the disease, and the family. People need professional help to understand what the result of a genetic test means for them.

1.10 Genetic factors also underlie many common diseases, including some of the country's biggest killers such as cancer, heart disease and diabetes. A substantial research effort is underway to investigate the genetic basis of these diseases, and important discoveries are already emerging.

1.11 Susceptibility to some rare subsets of common diseases, such as breast and colorectal cancer, can be caused by a single defective gene and have clear hereditary patterns. Where there is a significant family history, genetic testing can help predict the



chance of healthy family members developing the disease in the future. Monitoring and preventive regimes can then be offered to those at high risk. A negative test result can also reassure family members who do not carry the faulty gene. If symptoms are already apparent, genetic testing can identify the particular variant of the disease so that the most appropriate treatment can be chosen.

- 1.12** An example is the BRCA1 gene. About 1 in 800 women are thought to have inherited a defect in this gene. This means they have a lifetime risk of developing breast cancer of up to 85% and up to 60% for ovarian cancer. This is their inherent personal risk because of their DNA and is irrespective of other risk factors. Women with a strong family history of breast cancer are already being offered testing for defects in this gene. Those who test positive may be offered regular mammograms to detect any cancer at an early stage. They can also be prescribed a drug such as tamoxifen, or opt for prophylactic surgery, to reduce the risk.
- 1.13** It is likely that more of these familial subsets of common diseases will be discovered and tests developed for them.

Colon cancer

A patient today: At the age of 49, Jeff is diagnosed with advanced colon cancer. Even with radical surgery and chemotherapy, his chances of surviving to 50 are slim. Although devastated by the news, Jeff isn't completely surprised. His older sister Jean and his uncle died of bowel cancer, and looking back he realises that the abdominal discomfort and tiredness he has suffered intermittently for a year may have been early signs.

(continued overleaf)



Colon cancer – continued

In the future: When Jeff's sister Jean tells him that she has developed colon cancer, Jeff mentions this to his GP and is promptly referred to the primary care genetics counsellor. Jeff's family history of cancer is carefully pieced together, using the latest risk assessment software. At the counsellor's suggestion, Jeff and his son Steve each agree to have a genetic test. The test results show that Jeff has the same version of the gene that Jean has. Luckily Steve has a normal version and does not need to worry. Jeff's GP organises regular blood tests for him, to check for the first signs of any tumours. Five years later, when Jeff's blood test is found to be positive, he has a colonoscopy which confirms the presence of an early tumour. This is successfully treated without the need for radical surgery and he makes a good recovery.

- 1.14** Most of the more common diseases, such as heart disease and diabetes, also have a genetic component. An individual's susceptibility to these diseases is determined by a combination of genetic factors and environmental factors such as lifestyle and diet. The chances of someone developing a multifactorial disease are not nearly so clear-cut as with single gene disorders. A few tests already exist, but they have low predictive power and have not been used outside research settings. For example, a variation in the ApoE gene is associated with the development of Alzheimer's disease – but only a proportion of people with this mutation go on to develop Alzheimer's. And not everyone who develops Alzheimer's has this variation. This indicates that many factors are involved.
- 1.15** Over the next decade, however, it should be possible to identify more genetic factors that increase the likelihood of people developing a given disease. There will then be the option to test people for a predisposition to that disease, or a higher-than-normal risk. Preventive and monitoring services could then be tailored to an individual's needs.



- 1.16** Following on from this, the way external factors and genes interact to cause disease or protect us from disease will be better understood. This information will allow people with certain genetic profiles to avoid foods, chemicals or environmental factors, such as smoking, which are particularly risky for them.

Coronary heart disease

A patient today: Ali has a heart attack and is lucky to survive. After returning from hospital, his doctor explains that coronary heart disease (CHD), which can lead to heart attacks and strokes, is one of the commonest causes of premature death in the UK, but action can be taken to reduce the chances of another attack.

Ali is prescribed tablets for high blood pressure and high cholesterol. He resolves to try harder to lose weight, eat more healthily and take more exercise. Secretly, he wishes he had taken these things seriously before.

In the future: Primary care teams are able to identify people at genetic risk *before* they develop heart disease. When Ali moves house and registers at a new practice, the nurse uses a genetic test to assess his risk of CHD. Although Ali does not yet have any symptoms, and his blood pressure is normal, the test shows him to be at high risk because of his genetic make-up. Ali and his GP are then able to make more personalised decisions on lifestyle changes or drug therapy to reduce his likelihood of developing heart disease.

- 1.17** Research into the genetic mechanisms underlying disease will lead to more accurate diagnoses and more effective treatment. For example, rather than simply diagnosing that a patient has hypertension, doctors could, in the future, identify which of the underlying molecular mechanisms has gone wrong in a patient. Different treatments could then be recommended for different versions of a disease.



Diabetes

A patient now: Kate is 18 and has just started university. She should be making new friends and enjoying her studies, but instead she feels increasingly tired and lethargic. She is always thirsty, and starts to lose weight. Eventually she is diagnosed with diabetes, and is referred to a specialist team at the local hospital, where she is put on insulin treatment for life. Although she feels better, she will have to monitor her blood sugar levels from now on and inject herself with insulin every day.

In the future: Kate talks to her GP about the history of diabetes in her family and they decide she could have an inherited form of the disease. After discussion with a genetic specialist nurse she agrees to have a genetic test done and is told that she has Maturity Onset Diabetes of the Young. Unlike most cases of diabetes which develop in younger people, this unusual form of the disease does not always require insulin treatment and can be managed by Kate taking tablets and adjusting her diet. Kate is relieved that she does not have the worry of injections, and she soon feels well enough to enjoy university life to the full. Other family members, including her younger sister Anna, are counselled about their risk and offered genetic testing.

Pharmacogenetics

1.18 Variations in our genes mean that different people can respond differently to drugs. Some patients may not respond to a specific medicine, while others experience adverse reactions which can be serious. The science of pharmacogenetics looks at the underlying reasons for this, which can include:

- **genetic differences in drug metabolism:** Some patients metabolise certain medicines much more quickly than others, due to genetic differences in how their liver enzymes work. These patients will need a higher dose of the drug in order to obtain the blood concentrations necessary to work. Conversely, patients who metabolise more slowly might be at risk of accumulation of the drug and could experience toxic effects when given the normal dose. If



doctors, nurses and pharmacists had information about a patient's metabolic patterns, they could work out the most appropriate dose for them

- **idiosyncratic adverse reactions:** Other patients experience severe reactions such as hypersensitivity, regardless of dose. Certain genetic features seem to predispose patients to these severe reactions. Testing for these genetic features would make it possible to identify vulnerable patients and avoid giving them the medicine concerned
- **genetic predictors of response:** Some patients fail to respond to the medicine at all, perhaps because they have a variant of disease that is triggered by a different genetic mechanism to the one the medicine is designed for. Where these differences are affected by genetic variations, the patients who are likely to respond, or not respond, could be identified before treatment starts.

1.19 With greater understanding about how genes and drugs interact, many patients could undergo a genetic test to predict their response and help ensure the medicine and dose is right first time. This could:

- improve therapeutic outcomes and reduce the wastage involved in having to try out several products before finding the best one for each patient
- avoid serious and sometimes catastrophic side effects. Adverse drug reactions are estimated to affect around 7% of patients or hospital admissions at an annual cost of about £380 million to the NHS in England alone. This adds up to a huge burden on patients and on the healthcare system. Some of this is avoidable through better application of what we already know about medicines. But pharmacogenetics has the potential to significantly reduce the risk and cost of adverse drug reactions.

1.20 New pharmaceutical products linked to a pharmacogenetic test are likely to become available within the next five years. Where pharmacogenetic testing is shown to be important, the organisation of testing services, and the speed with which the result of the test can be known and acted on, will become key factors in helping patients receive the treatment that is best for them. New technology is currently being developed that should allow pharmacogenetic tests to be carried out quickly and cheaply in primary care settings.



Pharmacogenetics and warfarin treatment

A patient now: Mary has been feeling unwell recently, weak and shaky. At 58 years old she suspects it is just her age, but she goes for a check-up just in case. Her GP diagnoses a heart condition known as atrial fibrillation. This could cause Mary to have a stroke if small blood clots fly off the larger clot forming on the inside of her malfunctioning heart and travel through her blood to her brain.

After several unsuccessful attempts to treat Mary's condition, her GP prescribes warfarin to reduce the risk of a stroke. But it also increases the risk of excessive bleeding and bruising.

Mary's GP tells her what dose of warfarin to start taking but explains that she will need to have regular blood tests to measure the effect of warfarin on her blood. For the first month, tests will be required weekly to adjust the dose of the drug, and once it has stabilised, blood tests will be taken every 4-8 weeks.

Mary's sister is suddenly taken ill, and Mary misses her first few blood test appointments. She carries on taking her tablets at the dose her GP recommended, but she suddenly collapses on the way to the shops. In hospital, Mary discovers that she has had severe internal bleeding and was lucky to survive. While her initial warfarin dose was the normal starting dose, it was too high for her personally, and her blood was not clotting when it needed to.

In the future: Warfarin metabolism occurs at different rates in different people because of genetic variations in metabolism. It can also vary from day to day because of interactions with other medicines and other factors. Mary is one of the 4-5% of people who metabolise warfarin particularly slowly and therefore has a much higher risk of excessive bleeding and bruising, especially at the start of treatment. Mary's GP does a quick pharmacogenetic test using special equipment in the surgery. This enables her to identify Mary as a slow metaboliser and start her on a lower, safer, dose of warfarin that is better suited to Mary's personal genetic make-up.



Other uses of DNA analysis in healthcare

- 1.21** Genetic technologies are already widely used in pathology in areas such as microbiology and cancer. Genetic techniques can enable a much more detailed analysis of patients' cancer cells or of pathogens such as tuberculosis. This information can allow clinicians to predict with much greater accuracy which treatments will work.
- 1.22** In the next few years we can expect to see small scale technology able to carry out DNA analyses of viruses and microbial infections in primary care and emergency settings. Where a rapid result is required this could be preferable to sending a sample to a laboratory for analysis. For example it could allow a doctor or nurse to find out immediately whether someone with symptoms of meningitis had the disease or not, or whether a person was suffering from flu or just a cold.

Gene-based treatments

- 1.23** Greater understanding of human genetics will lead to a significant increase in the number of new drugs and novel therapies. It will allow the development of gene-based drugs and treatments targeted at the disease-causing fault rather than at the control of symptoms.
- 1.24** Unlike more conventional treatments, gene-based medicines work towards a direct interaction with the function of our genes within the body. For example gene-based medicines can aim to switch a helpful gene on or a harmful gene off. Such medicines are designed to work on our genetic material, DNA or the related molecule RNA. Gene therapy is the best known type of gene-based medicine.
- 1.25** Gene therapy is the deliberate introduction of genetic material into a patient's cells in order to treat or prevent a disease. As well as replacing defective copies of genes, a variety of sophisticated techniques are being developed that may allow the activity of genes to be turned up or down. Although this new branch of medicine is still at the research stage, we expect to see licensed gene therapy medicines within a decade. More than 600 trials have been approved world-wide and more than half of all European gene therapy clinical trials take place in the UK. These are targeting a wide range of conditions, from rare inherited disorders through to cardiovascular disease, cancers and infectious diseases such as HIV.



- 1.26** The full promise of gene therapy won't be known for at least another five years. At the current rate of development we can expect to see the first licensed gene therapy medicines coming on stream within five to ten years. In the longer term gene therapy may become a cornerstone of modern medicine.
- 1.27** Gene therapy offers hope to people suffering from those single gene disorders where there is currently no treatment at all, or where treatment options are very limited. It may also bring more patient friendly treatments for example in haemophilia, potentially replacing a lifetime of injections with a single treatment to correct the genetic defect which causes the disease.

Gene Therapy for Haemophilia A

A patient now: Wayne was diagnosed with haemophilia A when he was 6 months old. Haemophilia A is a blood clotting disorder caused by the disruption of a gene on the X-chromosome. Uncontrolled internal bleeding can occur in haemophiliacs. Wayne's disrupted version of the gene means his liver does not produce the Factor VIII protein which circulates in the blood, helping to make clots in wounds and limit bleeding.

Any minor knock can cause bleeding, and Wayne has to have clotting factor injections twice a week to try to preserve his joints from the crippling effect of bleeding as he grows through the rough and tumble of a normal childhood. He faces the possibility of a lifetime of injections, serious bleeds and added complications to any operations he might need.

In the future: Wayne's parents are offered the possibility of treating his haemophilia using gene therapy. They agree, and a healthy copy of the Factor VIII gene is transferred into Wayne's liver via gene therapy. This allows his liver to produce effective levels of the Factor VIII clotting agent on its own, without the need for twice weekly protein injections. This effectively cures his haemophilia and Wayne can lead a normal life without the need for further treatment.



1.28 Gene therapy also offers potential for the treatment of a wide range of common conditions such as cancer and coronary heart disease where its first uses will most likely be in the following areas:

- to kill cancerous cells more effectively and selectively and to increase the effectiveness of conventional cancer drugs
- to prevent and treat cancers by boosting the ability of the patients' own immune system to recognise and destroy tumour cells
- to treat coronary heart disease, by generating new blood vessels and improving blood flow around the heart.

1.29 The potential future impact of gene therapy is enormous. Cancer is the cause of a quarter of all deaths in the UK. Cancer causes an even greater proportion of deaths in those aged under 65, where more than one in three deaths is due to cancer. Gene therapy trials are ongoing for most of the big killers:

- Colorectal cancer – the most common cause of cancer related deaths in the UK. 28,000 new cases and 19,000 deaths a year
- Breast cancer – the most common cause of death in women between the ages of 35 and 54 years. Around 34,000 new cases a year
- Ovarian cancer – a major cause of death for women with an estimated 4,500 deaths a year
- Prostate cancer – around 20,000 new cancers diagnosed each year
- Brain tumours – 4,000-5,000 patients are diagnosed with brain tumours each year.

1.30 Although early days, gene therapy trials for coronary heart disease are also showing encouraging results. CHD is the commonest cause of death in the UK. One in four men and one in five women die from CHD.

1.31 The first gene therapy clinical trials took place in 1990. Although there have been some marked successes and some very promising results in clinical trials, gene-based medicines are unlikely to have a significant impact on the lives of many people for at



least a decade. However, some experts predict that by then we will see approved treatments for a range of diseases, not only in the major areas of cancer and cardiovascular diseases, but also promising developments in areas as diverse as arthritis and some forms of blindness such as macular degeneration which accounts for almost 50% of blind registrations in the UK.

- 1.32** A new approach known as RNA interference (RNAi) is thought to hold great promise for the future of gene therapy. It is being used by scientists in the laboratory to deactivate specific genes in living cells, in order to learn more about their functions. In future, it may be possible to use RNAi to disable specific genes which cause disease in patients.
- 1.33** Good progress is also being made in developing gene based vaccinations, although marketed products are likely to be several years away. Trials of cancer vaccines have already started. The vaccination stimulates the immune system to recognise and destroy the tumour cells, helping the patient's body to fight the disease. In the future, such vaccinations might also be developed for those at high risk of cancer to help prevent tumours from occurring in the first place. Gene based vaccines are also being developed against a number of difficult to treat infectious diseases including hepatitis B, hepatitis C and HIV.

Realising the benefits

- 1.34** The new genetics knowledge and technology has the potential to bring enormous benefits for patients: more personalised prediction of risk, more accurate diagnosis, safer use of medicines and new treatment options. A revolution in healthcare is possible, but it will not happen overnight.
- 1.35** The Government has an ambitious vision for harnessing these potential benefits for patients. We want the NHS to lead the world in taking maximum advantage of the safe, effective and ethical application of the new genetic knowledge and technologies as soon as they become available.
- 1.37** Our vision for the NHS needs an effective delivery strategy. We intend to support specialised services and to encourage and assist clinicians and managers throughout the NHS to incorporate genetic advances into everyday clinical practice.



- 1.37** The exact timing for different genetic advances is uncertain and it is too early to accurately predict all our requirements for the next decade. But unless we act now to ensure we have firm foundations to build on, the NHS will be left trailing. To make a reality of our vision this White Paper sets out a detailed plan of action and investment for the next three years. We will review progress at the end of that time to see what more needs to be done.
- 1.38** We will help the NHS lay the foundations for success by:
- providing investment to strengthen existing hubs of NHS expertise. We will boost capacity within specialised genetics services by continuing to expand the workforce and we will earmark substantial capital investment for a major programme of modernisation for genetics laboratories (**chapter two**)
 - assisting genetics to permeate all branches of medicine by supporting new initiatives in genetics based care in key diseases areas, in secondary and primary care, and through national screening programmes (**chapter three**)
 - helping to empower frontline staff and commissioners in the NHS through investment in education and training, information systems and developing the evidence base (**chapter four**)
 - generating new knowledge and applications through investment in genetic research and development (**chapter five**).
- 1.39** We want to ensure that patients across the country benefit from the advances in diagnosis, prevention and treatment that genetics will bring. At the same time we recognise that advances in genetics raise difficult ethical and social issues. We intend to make sure that these are widely discussed in an open and transparent way and that we receive independent and informed advice. Where necessary we will take action to introduce appropriate controls. We are committed to increasing public understanding of genetics and ensuring public confidence through a robust and proportionate system of regulation (**chapter six**).

2. Strengthening specialised services

2.1 There are 18 specialist NHS genetics centres in England. They bring together clinical geneticists, genetic counsellors and genetics laboratories and they are highly regarded across Europe for their expertise and leading edge practice. These centres:

- use a combination of clinical examination, family history taking and laboratory tests to make accurate diagnoses of genetic disease. They explain what the diagnosis means and advise on medical problems connected with the genetic condition
- advise people with a strong family history of disease about their own chances of developing the condition or passing it to a child
- help people with a family history of late onset disease such as Huntington's to decide whether or not to have a genetic test.

2.2 Clinical geneticists are increasingly working with other medical specialists as the relevance of genetics to their clinical fields becomes apparent. The work of laboratory scientists is also diversifying as new genetic tests become available and demand for their services grows.

2.3 These specialist centres have a vital role in spreading genetic expertise across the NHS. With this in mind, the Department of Health has already committed more than £11 million to boost the capacity of clinical and laboratory genetics.

2.4 The two biggest constraints in developing the specialist hubs are the workforce and infrastructure. **So the Government will provide:**

- **new investment to expand the specialist genetics workforce**
- **a major programme of new investment to modernise the genetics laboratory service**
- **new investment to support information systems in genetic centres.**



Investing in the specialist genetics workforce

- 2.5** The NHS Plan recognised the shortage of human resources as the biggest constraint faced by the National Health Service. It set out commitments to increase workforce numbers, and led to the setting-up of a new structure for workforce planning for the NHS.
- 2.6** Numbers of specialist genetics staff will need to grow to ensure that NHS services are prepared for patient demand. Laboratory staff will be needed to provide for the increasing volume of genetic testing. Consultants and counsellors will be increasingly called upon to advise and liaise with other clinical colleagues.
- 2.7** The Department has already provided start-up funding in the past year for an additional 11 genetics consultants, 28 genetics nurses and counsellors and 42 genetics laboratory staff. Commissioners will need to pick up these costs from 2004-5 aided by the large increases in NHS expenditure that this Government is providing over the next five years.
- 2.8** Increases in the number of training places available are already helping to boost numbers of genetics consultants. Central funding was provided to support the implementation of eight additional specialist registrar posts in clinical genetics in 2002/03 and a further ten additional posts in 2003/04. Trusts will also have the opportunity to create up to five locally funded SpR training opportunities.
- 2.9** There are currently around 200 genetic counsellors in England. Increasing training capacity is a priority. **The Department of Health will fund a new scheme of new clinical training posts to train over 50 new genetics counsellors** over the next five years. These will be based in specialist genetics centres and will provide a new route to qualification as a genetic counsellor. The Department of Health is also funding additional places on the vocational MSc courses in genetic counselling in Manchester and Cardiff and has provided funding for clinical placements in these and other centres.
- 2.10** Genetic counselling is an emerging profession and the Department of Health is supporting moves to give these practitioners a strong professional identity. The Association of Genetic Nurses and Counsellors has devised a professional registration process and it is now registering those who have the required qualifications and experience.



- 2.11** Over the coming years, more genetics laboratory staff will also be needed to meet the rising demand for genetic testing. **The Department of Health will make available up to £3.5 million for a strategic programme to boost the workforce in genetics laboratories. This money will fund up to 90 new Grade A trainees in laboratory genetics, more than doubling the present capacity.**
- 2.12** Genetics laboratories can often recruit staff who need training, but experienced staff find it less easy to release time from existing commitments to provide that tuition. **The £3.5 million announced above will also be used to fund the equivalent of ten full time trainer posts. This will allow experienced staff to be released from service commitments to provide training. It will also fund some nationally based training for new trainees and existing staff.**
- 2.13** This strategic investment will greatly strengthen training capacity for the long term. But more immediately it will provide a significant cohort of new qualified staff. Other moves will also enable staff in genetics laboratories to be better placed to meet future challenges. New national occupational standards are being developed for clinical molecular genetics and clinical cytogenetics. These will augment existing training programmes, leading to professional qualifications in healthcare science and will inform continuing personal development programmes at all levels of practice. Implementation of *Making the Change*, the Government's strategy for healthcare scientists, and successful implementation of *Improving Working Lives* for all staff will result in a workforce better equipped to deal with the increasing workload faced by genetics laboratories.

Investing to modernise the genetics laboratory service

- 2.14** Genetic testing will expand rapidly in the short and medium term. As more genes are discovered, sequenced and their functions determined, so the ability to test for abnormalities will grow. Over time, the range of tests and the number of tests carried out will increase dramatically.
- 2.15** This increase will be fuelled in part by growing consumer demand. As people become more aware of the part played by genes in ill health, they will naturally ask their GPs for genetic tests. There may be awareness raising campaigns by medical charities and patient support groups which draw attention to new genetic tests that



can be requested from NHS doctors. Whether or not commercial companies should be able to market tests direct to the public is considered in chapter six. But if genetic tests do become available over the counter this could further increase demand on the NHS as patients, and their GPs, may well want to validate an adverse test result from a privately purchased genetic test.

- 2.16** To prepare the NHS genetics laboratory service for these challenges, the Government will provide a major programme of new investment so that facilities can be upgraded and capacity increased. A radical reshaping of the service may also be necessary to maximise the benefits of more centralised working, closer links with pathology or partnership with commercial laboratories.

The challenges ahead

- 2.17** Until now, genetic testing has been used mainly for single gene disorders. However this is changing. Genetic testing for breast and bowel cancer has increased markedly. The discovery of more familial subsets in cancer and other diseases will further increase the volume of tests. And as genetic testing becomes relevant for more common diseases, the number of people to be tested could be very much larger still.
- 2.18** Predisposition and pharmacogenetic tests are likely to involve screening for small variations in DNA sequence, known as single nucleotide polymorphisms (SNPs). These are simple to test for, but there will be millions of these possible genetic variations, often needing to be tested in combination. This could have a profound impact on the laboratory service.
- 2.19** Newer genetic techniques are enabling genetics laboratories to provide very rapid results in some areas of work, notably in antenatal diagnosis and screening programmes. This will be an increasing feature of genetic testing in the future. Results for some newer types of tests, such as cancer typing or pharmacogenetics, will often be needed within hours to inform urgent treatment decisions.
- 2.20** High-throughput automated testing technology, often operated round the clock, will become essential to deliver quality results quickly. Robotics and other automation will mean that testing at vastly increased speeds and the ability to do more tests at once will be possible, and the cost per test reduced considerably.



- 2.21** Near-patient testing technology offers scope for some types of genetic tests to be carried out at the patient's hospital bedside or in GP surgeries or pharmacies. The possibility of almost instant results could well tip the balance in favour of using genetic tests in new situations. This new technology is being developed now and may be marketed within a few years.
- 2.22** The prospect of cheap whole-genome scanning could bring entirely new opportunities. In theory, a patient's whole genome could be scanned once and the results interrogated later. The need to store and interpret such vast quantities of computerised data will produce real challenges in bioinformatics.

Progress to date

- 2.23** Progress has already been made in preparing NHS genetics laboratory services for the future:
- much of the £11 million committed by the Department of Health to genetics services in England over the last year has been channelled into laboratories, to increase their capacity and reduce the time patients have to wait for results
 - building on long-standing professional collaboration, the different genetic centres in the UK have formed the UK Genetic Testing Network. This will facilitate co-operation and work-sharing between different laboratories
 - two National Genetics Reference Laboratories funded by the Department of Health have been established to support the UK Genetic Testing Network.



National Genetics Reference Laboratories

Two National Genetics Reference Laboratories were set up in 2002 in Manchester and Salisbury. Their role is to support other NHS genetics laboratories in this rapidly developing field, so that patients can benefit from the best testing service possible. The Reference Laboratories are:

- developing new ways of testing
- researching and evaluating new technologies
- offering training
- expanding existing quality assurance programmes
- disseminating information on best practice

2.24 However there is still much to be done. Many NHS genetics laboratories have struggled to achieve the space, workforce and equipment they need. While many laboratories have achieved accreditation, others have yet to meet this standard. **The Government now expects all genetics laboratories offering clinical testing to secure accreditation with CPA (Clinical Pathology Accreditation Ltd) or equivalent within 18 months.** This will reassure patients, clinicians and commissioners about the quality of work.

Investing for the future

2.25 To cope with the expected increase in genetic testing, the Government will make a substantial investment in NHS genetic testing capability. We want to increase capacity, take advantage of new technology and testing methods, maintain rigorous quality standards, and offer flexibility to adjust to changes in the focus of genetic testing. We will set up a strategic investment programme to help genetics laboratories achieve these aims. **We will invest up to £18 million capital during the period 2003-6 on upgrading NHS genetics laboratory facilities in England.** We will also provide some start-up funding for new staff and consumables.

2.26 This unprecedented level of investment provides an opportunity to make sure that genetic testing services are organised in the most effective way. It will put



laboratories on a sound footing for the future and it will benefit patients in the short term by improving access and cutting waiting times for test results. **As a result of this investment, by 2006 genetic test results should be available to the following standards:**

- **within three days where the result is needed urgently (e.g. for prenatal diagnosis)**
- **within two weeks where the potential genetic mutation is already known (eg. because another family member has already been tested)**
- **within eight weeks for unknown mutations in a large gene.**

Configuration of genetic testing services

- 2.27** For some time, there have been calls to re-examine how the genetics laboratory service is best configured. In August 2000 an expert working group published a report on *Laboratory Services for Genetics* and recommended no immediate changes to the existing configuration of specialist genetic laboratories, but said this must be kept under review.
- 2.28** The Pathology Modernisation Programme suggests that there can be significant advantages in centralising smaller services or organising laboratory services more coherently across a geographical area. Quality and efficiency have been improved, allowing laboratories better to support and improve clinical decision-making. Guidance for the NHS on modernising pathology services is being developed and is due to be published this summer.
- 2.29** It is now time to take a fundamental look at how genetic testing is organised. Radical changes will have to be considered if genetic testing services are to cope with the expected growth in demand. Questions to be addressed include:
- how many genetics laboratories are needed to serve a given population
 - whether all genetics laboratories need to undertake all types of test or whether there is scope to distribute work between laboratories more effectively



- whether there should be closer links, or even mergers, between clinical genetics laboratories and pathology laboratories which are increasingly using DNA techniques
- what role the private sector can play.

2.30 In considering the desirability of these options, a range of factors including costs and practicality will have to be taken into account. Greater integration between laboratories or centralising work could:

- produce greater economies of scale
- make more efficient use of expensive equipment, perhaps running over a longer working day and serving multiple uses, or providing a specific service for a larger geographical area
- make better use of skilled staff, provide additional career opportunities and allow more flexible working patterns
- provide a critical mass of tests that would allow laboratories to maintain quality standards and professional expertise more easily.

2.31 Any new configuration must be sufficiently flexible to cope with future requirements. The exact shape, scale and timing of future demand will be difficult to predict, and much depends on how technology develops. Other factors might affect the cost, practicality or desirability of these options:

- moving some genetic testing workload off-site could have implications for timeliness because of the additional time needed to transport samples, or quality if on-site clinical involvement is needed in the interpretation of some results and an IT solution cannot be found
- redistributing work could have wider consequences for laboratories. For example, if the range of tests offered is significantly reduced, it could limit a laboratory's ability to attract new staff and to provide comprehensive training for them
- arrangements involving the private sector would need to be of interest to commercial partners as well as offering the NHS value for money and quality.



Robust safeguards to ensure the confidentiality of patient data will also be vital as well as careful agreement over the use a private company is allowed to make of the data.

Assessing local options for strategic investment

2.32 We will invite bids from commissioners and providers. Drawing on experience from the Pathology Modernisation Programme, they will need to take an in-depth strategic look at how their services might best be organised to increase capacity and maximise quality, considering both current workload patterns and potential future demand. We will look closely at how proposed schemes will achieve this and will expect bidders to have considered the following options:

- setting up an integrated genetics and pathology laboratory service
- amalgamating or substantially reconfiguring services across two or more existing genetics laboratories within an area
- substantially reconfiguring working practices through skill mix or robotics or new IT solutions
- contracting out suitable work
- including the private sector as partners.

2.33 Involving the private sector could bring valuable benefits. Contracting out work to commercial laboratories might allow the NHS to concentrate on more specialist areas. Capital from the private sector could make more ambitious projects possible, for example by part-funding new laboratory facilities and equipment. Partnerships between the NHS and private sector could provide the expertise and new technologies to organise and carry out work more efficiently.

2.34 It will take time and expertise to prepare the high quality bids we expect. Bidders will need to demonstrate that they have considered all the options, have the full involvement and ownership of staff, and have drawn on the views of key stakeholders including patients. To support this process we will:

- give a six-month period for bids to be prepared



- host one or more workshops where commissioners and service providers can share thinking, learn from earlier experiences and perhaps collaborate
- offer a small amount of funding to support the costs of preparing the bid including the local appraisal of options.

2.35 Drawing on appropriate external expertise, the Department of Health will assess the bids against open and transparent criteria, in particular making sure that the selected bids contribute to a coherent national picture.

Investing in information systems

2.36 Specialised genetics services are already taking steps to modernise and harmonise their IT systems. Some centres are transferring patient records onto electronic format and linking laboratory data into these. The National Genetics Reference Laboratory in Manchester and the Clinical Molecular Genetics Society have convened a working group to identify lessons from current initiatives that could assist the UK Genetic Testing Network. Examples of projects which are already delivering local benefits are:

- a portal system developed by St George's Hospital, and linked to several other genetic centres in London, which allows genetic tests to be requested and the results retrieved electronically
- an information management system developed in Newcastle to improve information returns to those requesting the tests
- a design specification for laboratory and information management systems being developed in Manchester for handling and tracking patient samples through the system.

2.37 All these moves will reduce paperwork and the scope for error, allow for greater integration, and result in better management of workload and patient information in individual laboratories. However in order to maximise the efficient functioning of the Genetic Testing Network, **all** participating laboratories will need to work with compatible systems that would:

- allow samples to be tracked throughout the Network



- facilitate communication and linkage between laboratories
- provide better information for workload and quality management, including support for audit and validation of laboratory data
- improve the quality, efficiency and timeliness of reporting results for patients.

2.38 To support these developments, **the Department of Health will invest up to £1 million over three years in IT for genetics laboratories in the Genetic Testing Network. This will enable better handling of work and communications within and between participating laboratories.** These new systems will be compatible with the Government's wider investment programme to modernise NHS IT systems and introduce electronic patient records.

Conclusion

2.39 The Government has an ambitious vision for the NHS to be the world leader in applying the benefits of the new genetic knowledge in healthcare. The critical first step is to develop existing centres of expertise and overcome the twin constraints of inadequate staff numbers and infrastructure.

2.40 The Government will:

- we have committed over £11 million to specialised genetics services over the past year and we have made a start in expanding the genetics workforce
- we have set up new Genetics Reference Laboratories in Manchester and Salisbury and established a UK Genetic Testing Network
- we will continue to invest in expanding the specialist genetics workforce. In particular, we will fund new clinical training posts to train over 50 new genetics counsellors over the next five years. We will also make available up to £3.5 million for a strategic programme to boost the workforce in genetics laboratories. This money will fund up to 90 new Grade A trainees in laboratory genetics, more than doubling the present capacity, and the equivalent of ten full-time trainer posts



- **we will invest up to £18 million capital during the period 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 of this investment, by 2006 genetic testing times will be cut and the results should be available to the following standards:**
 - **within three days where the result is needed urgently (e.g. for prenatal diagnosis)**
 - **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.**
- **we will invest up to £1 million over three years in IT for genetics laboratories in the Genetic Testing Network. This will enable better handling of work and communications within and between participating laboratories.**

3. Building genetics into mainstream services

- 3.1** Strengthening existing specialised services is an essential first step. Modernised laboratories will also provide the essential infrastructure to meet the increased demands for patient testing.
- 3.2** Over time, as the relevance of genetics across the spectrum of other medical specialties becomes apparent, mainstream NHS services will be able to take advantage of new genetic tools in diagnosing, preventing and treating disease. Genetic technologies should increasingly help clinicians to identify people at risk of disease and to provide their patients with more personalised care and treatment which suits their genetic profile.
- 3.3** The specialist genetics centres will play a leading role in spearheading the diffusion of new genetic advances across the rest of the NHS. The Government will also spur the take-up of genetics by other specialties by supporting new initiatives in genetics based care – in the hospital sector, primary care and in screening programmes.

Engaging other specialties

- 3.4** Genetics will become increasingly relevant to diseases such as cancer, heart disease and diabetes, which are priority clinical areas. Researchers are also exploring genetic influences in asthma, osteoporosis, mental illnesses such as schizophrenia and many other conditions.

Cancer

- 3.5** A small proportion of breast, ovarian and bowel cancers are associated with particular inherited genes. Testing for these genes can identify whether someone with a strong



family history of the disease is likely to develop it and ensure they have regular checks or treatment. In the future, more of these familial subsets of cancer may be discovered, for example in skin cancer. It may also become possible to test for genes or combinations of genes that predispose to cancer in a less clear-cut way, for example by increasing susceptibility to harmful environmental stimuli such as cigarette smoke.

3.6 This area of clinical and laboratory work is expanding as awareness of inherited cancers grows. Some cancer specialists are working closely with geneticists and GPs to decide on the most effective way to identify and manage people at different risk of developing familial cancers. The National Institute for Clinical Excellence is also preparing clinical guidelines on the identification and management of women who have inherited genes that predispose them to breast cancer. Publication is expected in February 2004.

3.7 Macmillan Cancer Relief, working closely with the Department of Health, service users and leading experts in cancer and genetics, have developed a model approach to services for people at risk of, or concerned about, familial cancer. The model offers a continuum of advice and care involving primary care, local cancer services and specialised genetic and cancer services. It includes:

- the provision of consistent, correct and appropriate information for service users
- risk assessment according to an agreed national framework
- streamlined referral in accordance with agreed pathways
- consistent management of individuals in the appropriate setting according to their level of risk.

3.8 **Macmillan are now ready to pilot this model. The Department of Health will co-fund this initiative and provide sufficient resources to pilot the model in six cancer network areas. We will also provide funding for an evaluation of the lessons learned and the implications for a national programme.**

Heart disease

3.9 Heart disease is known to run in families. There are genes that can increase the risk of high blood pressure, high cholesterol or clotting abnormalities. Researchers are



currently trying to learn more about the genetic causes of heart disease. We need a better understanding about the effects of single genes and multiple genes working together to be able to predict the risk of heart disease developing in an individual.

- 3.10** One example is familial hypercholesterolaemia. This is an inherited disorder giving rise to high blood cholesterol levels and a significantly increased risk of heart attacks at an early age. Around 1 in 500 of the population have this defective gene, or about 100,000 people in England. Treatment with drugs to lower cholesterol is effective in reducing disease and death amongst this group.
- 3.11** There is also good evidence to support a programme to identify people with familial hypercholesterolaemia and to offer them treatment **before** symptoms develop. Cascading the offer of a test to close family members (parents, brothers, sisters and children) of people already diagnosed with familial hypercholesterolaemia has been successful in the Netherlands as a way of finding more people with the condition. In the first five years of a nationally funded programme, about 30% of all people expected to have familial hypercholesterolaemia have been identified and offered treatment.
- 3.12** This programme offers a paradigm for how to use genetics to improve healthcare outcomes for a complex disorder. **The Department of Health will fund a two year programme of pilots around the country to identify and treat people with familial hypercholesterolaemia in order to reduce their risk of dying of a heart attack at an early age.** The pilot programme, based in five lipid centres around the country will identify at least 1,000 new patients. It will also allow us to carry out research to resolve remaining questions about the ideal approach and to learn important lessons for an effective national programme.

Diabetes mellitus

- 3.13** A family history of diabetes is known to increase the risk of developing diabetes. Apart from one form of diabetes known as maturity onset diabetes of the young (MODY), the precise gene or genes involved in diabetes are not yet fully understood.
- 3.14** The Department is funding a project based in Exeter to educate specialist diabetes nurses around the country to help spot patients with MODY. Differentiating these



patients can help ensure they receive the right treatment for this particular form of diabetes. It can also be useful in establishing the implications for their families.

- 3.15** As evidence emerges across all clinical areas, genetic knowledge and technologies will need to be taken into account in the work programme for NICE and inform the development of National Service Frameworks. These will continue to evolve as new evidence is available and as we learn through implementation.

Fostering new initiatives

- 3.16** **The Department of Health will invite bids for up to £2 million of start-up funding over three years for initiatives to bring the benefits of genetics into mainstream clinical areas.** We would like to encourage local innovation and good ideas and bids might include:

- using the skills of specialist nurses, counsellors or other health staff to help patients explore their family history or advise them about their genetic risk. They could also act as a link between the different specialties and a common point of contact for patients so that their care is properly co-ordinated
- joint clinics, perhaps using telemedicine or other innovative methods, to bring expertise from different specialities or professions together and provide a ‘one-stop shop’ for patients
- different specialities collaborating to produce and implement referral pathways or good practice guidelines, with designated roles and responsibilities for the different health professionals involved in patients’ care.

- 3.17** We will fund a wide range of ideas in many parts of the NHS, particularly proposals linked to the National Service Framework areas. This will help to kick-start the integration of genetic health care into other services, and provide practical experience for developing services in the future.

New investment in primary care

- 3.18** GPs, practice nurses and other primary care practitioners will all be able to help their patients benefit from the new genetic knowledge and its applications. They already



understand the long term, psychosocial aspects of illness. They work with individuals in the context of their families over time. They are adept at identifying health problems and making appropriate referrals. They co-ordinate the care of the affected patient. And they are at the forefront of health promotion and prevention.

Roles for primary care in genetics

- Managing patients' concerns and expectations
- Identifying genetic conditions
- Assessing risk
- Managing risk
- Screening
- Testing
- Providing and co-ordinating long-term care
- Gatekeeping to specialist care

- 3.19** As genetic testing technology becomes simpler and faster, common tasks in primary care such as the prevention of CHD and the management of asthma or diabetes could involve the carrying out and interpretation of genetic tests. Information from such tests could provide clinicians and patients with more accurate, personalised information about their health problems.
- 3.20** Pharmacogenetic tests could make the use of some medication safer and more effective by allowing the choice and dose of medicine to be tailored to an individual's personal genetic make-up. Warfarin, asthma drugs and anti-psychotic medication are potential early candidates. The logistics and clinical utility of including a test in prescribing decisions will need careful evaluation, but applying this knowledge within primary care should significantly improve patient outcomes in medicines use.
- 3.21** Developments in pharmacogenetics could allow pharmacists with specific training to take on a greater role in prescribing, working in partnership with GPs and nurses. A GP could make the diagnosis and discuss with the patient the general approach to



treatment. But the selection of the genetically most favourable option could be undertaken by a pharmacist with the aid of genetic testing facilities.

- 3.22** Some doctors, practice nurses or pharmacists may want to take a special interest in genetics. For example nurses may wish to extend their expertise by working specifically with families who are at particular risk of genetic disease. As well as helping to screen family members, they could, with appropriate training and support, advise doctors about high-risk patients who should be referred for further tests. This work has been begun with familial breast cancer, and in time, a role for locally based practitioners working across practices to help with referral and counselling may emerge.
- 3.23** The Department of Health is already funding three development projects in Southampton, Nottingham and West Hull which look at the role of genetic nurse counsellors in general practice.
- 3.24** Developing services in primary care will require investment in primary care staff and infrastructure. Primary care professionals will want sufficient time to develop new knowledge and to counsel patients and families properly. They will also want to understand the issues involved in testing and in recording genetic information in patients' medical records.
- 3.25** To help address these concerns and explore new ways of building expertise and capacity specifically in primary care, the **Department of Health will invite bids for up to £2 million of start-up funding over three years for primary care genetics initiatives.** This investment will be in addition to the enhanced services funding set out in the PCT allocations circular. It is important that these ideas should be developed locally and we anticipate a wide range of imaginative bids from individual PMS or GMS practices or groups of practices, or PCTs. These might include:
- using genetics nurse counsellors in primary care, employed by one practice or several, or on an outreach basis from the specialist genetics centre
 - establishing and promoting the development of good practice, for example in assessing genetic risk or making appropriate referrals, or testing how methods of support such as clinical networks can improve services
 - establishing lead clinicians in PCTs to shape new local services in genetics.



- 3.26** General practitioners with special interests supplement their important generalist role by delivering a high quality, improved access service to meet the needs of a single PCT or group of PCTs. They take referrals from their colleagues and they may deliver a clinical service beyond the scope of general practice, undertake advanced procedures or develop services. Progress to date suggests that the introduction of GPs with special interests bring with them real and sustainable benefits for patients and the NHS. They are providing localised services, in familiar surroundings with easier access and speedier care for patients.
- 3.27** There are already GPs with a special interest in a range of clinical conditions such as diabetes, dermatology and musculoskeletal medicine. **We would like to see the introduction of GPs with a special interest in genetics and the Department of Health will provide start-up funding for up to ten to be established over the next three years.**

Screening programmes for genetic disorders

- 3.28** National screening programmes aim to reduce disease by identifying people most at risk. But screening is not a perfect science and can cause anxiety. Some people may be wrongly reassured by a false negative result, and others with a false positive could undergo unnecessary follow-up testing. Screening large numbers of the well to identify the few at risk may not always be the best use of NHS resources. Because of these issues, new screening programmes must be approved by the UK National Screening Committee and endorsed by Ministers.
- 3.29** Screening programmes already exist for a number of disorders that have a genetic component. These are being extended to cover some additional conditions. **Two programmes, expected to be fully implemented in England by 2004/5, will ensure that:**
- **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. Some of the hearing problems picked up will have a genetic cause.**
- 3.30** Work is also in hand to offer cystic fibrosis screening to all newborn babies. They are already offered screening through the national blood spot programme for the genetic condition PKU (phenylketonuria), an enzyme deficiency which causes severe learning difficulties, and congenital hypothyroidism, which includes some inherited cases. If the baby is found to have PKU or hypothyroidism, the adverse effects can then be prevented by early treatment.
- 3.31** Screening programmes for the inherited blood disorders, sickle cell and thalassaemia are also being developed. In England **by the end of 2004:**
- **A newborn screening programme will be in place offering screening for sickle cell disease. This will cover around 320,000 births per year and pick up around 90% of affected infants**
 - **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.**
- 3.32** As new tests are developed and more knowledge about the genetic basis of disease is acquired, it will become possible to screen for more genetic disorders. Even a small advance could suddenly make a potential screening programme therapeutically worthwhile or cost-effective. For example, gene chips are being developed which can test for a number of genetic mutations at the same time.
- 3.33** The benefits could include earlier treatment of disease, predicting the risk of disease later in life, detecting carrier status and enabling more informed reproductive choice.
- 3.34** But there are also arguments against. Telling people they carry a genetic feature which they could pass on to future children may cause considerable worry or stigmatisation. Individuals who choose to be screened may also uncover risk implications for relatives who would have preferred not to know. To help address these concerns, representatives of the genetics community have drawn up a set of guiding principles to cover screening programmes that include a test for an inherited



genetic disorder. The National Screening Committee has agreed to update their existing criteria to reflect these principles. They include the need to consider:

- the implications for members of the wider family
- how to handle technologies that may provide results on more conditions than those in the screening programme. People need to know exactly what they are being screened for before they agree to take part. If there is a possibility that the technology will generate results about other conditions or carrier status, then the decision about what to do with this additional information needs to be carefully thought through before the screening programme is established. Where these additional results may have implications for a person's health, or the health of their children, it is ethically unacceptable to withhold that information unless specifically requested by that person
- the importance of identifying and resolving any ethical issues such as confidentiality of information, privacy, obtaining valid consent and storage and protection of genetic material.

3.35 Genetics will bring new challenges as well as opportunities for screening programmes. In time it may become possible to use genetic screening to identify people who are at higher than normal risk of developing common multifactorial diseases like heart disease or cancer. However it is not yet clear what benefits will come from warning people who are at higher genetic risk of disease. We do not know whether this would make them more likely to change their lifestyle or take other preventive measures. It may even have the opposite effect if people adopt a fatalistic approach and assume they're going to develop the disease anyway so there's no point in taking preventive action. And of course people whose test is clear may be more inclined to indulge in risky behaviour such as smoking as a result. Research will be needed to answer these and other questions so it will be many years before we know whether genetic screening programmes for multifactorial diseases are worthwhile.

Genetic profiling at birth

3.36 One long term possibility that has been suggested is to screen babies at birth as part of the standard postnatal checks and to produce a comprehensive map of their key



genetic markers, or even their entire genome. Major investment is currently being made in information technology in the NHS, including the development of an electronic patient record for each person. The baby's genetic information could be securely stored on their electronic patient record for future use. It could then be used throughout their lifetime to tailor prevention and treatment regimes to their needs as further knowledge becomes available about how our genes affect our risk of disease and our response to medicines.

- 3.37** However, although the technology may be with us to do this and although the health benefits could be great once genetic factors with sufficient predictive power are identified, such a proposal clearly raises a wide range of ethical and social concerns which would need to be fully explored. It would be essential that the genetic information was securely stored and access to it protected through statutory safeguards. Participation would be voluntary and individuals would need the right to withdraw their information at any time.
- 3.38** A more thorough knowledge about the meaning of genetic variations, together with public debate on many of the wider issues, is essential before we could consider introducing such a system. Although its possible introduction is some years away we believe it is important to start thinking through such possibilities now in order to make sure that we are best placed to capitalise on the health benefits in future, should it be widely agreed that the benefits outweigh the risks and that the risks can be satisfactorily avoided. **As a first step, we will ask the Human Genetics Commission to work with the National Screening Committee and conduct an initial analysis of the ethical, social, scientific, economic, and practical considerations of genetic profiling at birth and to report by the end of 2004.**

Conclusion

- 3.39** The Government will provide direct investment to support the spread of genetics into mainstream NHS services. We will:
- **co-fund with Macmillan Cancer Relief pilots in six cancer network areas of a model of delivering services for people at risk of, or concerned about, familial cancer**



- **pilot a systematic programme to identify and treat people with a genetic disorder known as familial hypercholesterolaemia in order to reduce their risk of dying of a heart attack at an early age**
- **provide up to £2 million of start-up funding over three years for other initiatives to bring the benefits of genetics into mainstream clinical areas**
- **provide up to £2 million of start-up funding over three years specifically for primary care genetics initiatives**
- **provide start-up funding to allow up to ten GPs with a special interest in genetics to be established over the next three years**
- **ensure that by 2004/5, all pregnant women are offered antenatal screening for Down's syndrome and then counselled by midwives to help them make an informed choice**
- **ask the Human Genetics Commission to work with the National Screening Committee to consider the case for screening babies at birth and storing information about their genetic profile for future use in tailoring healthcare according to their needs and their genetic make-up. The HGC will be asked to report by the end of 2004.**

4. Spreading knowledge across the NHS

- 4.1** The Government can help lay the foundations for genetics to be woven into NHS delivery by helping to spread knowledge among health care staff. We will:
- invest in education and training
 - support evidence based care
 - invest in information systems
 - support commissioners.

Education and training

- 4.2** Over time most healthcare professionals will need to understand how a patient's family history and genetic make-up affects their likelihood of developing a disease or their response to medicines. They will need an appreciation of how genetic technology can be used in diagnosis, prevention and treatment. And they will need to be able to convey this information to patients and help them make difficult choices about whether to undergo a genetic test or to change their lifestyle in the light of information about their genetic make-up.
- 4.3** Patients themselves will become more aware of the role of genetics and will increasingly start asking questions about familial risk, genetic tests and new therapies.
- 4.4** At the moment, provision and coverage of genetics within training courses is patchy. For most professions and specialities, including primary care, it will need to be developed and integrated into undergraduate and pre-specialisation courses. New



practitioners need to start their careers with a strong and up-to-date grounding in genetics.

- 4.5** Continuing professional education will also be vital. An important first step is to raise awareness in all clinical groups of the potential and relevance of genetics in their field of practice.

Progress to date

- 4.6** A number of education-related initiatives have already begun to take genetics into new areas:

Activity to date

Scoping and strategy project

- The Wellcome Trust and the Department of Health are co-funding a study on genetics education and training across all the main non-genetic clinical professions (medical practitioners, nurses, midwives, health visitors, pharmacists, dieticians) and health service managers. This is being undertaken by the Public Health Genetics Unit which is based in the Cambridge Genetics Knowledge Park.
- In Phase 1, the extent and content of genetic education and training within current educational programmes was mapped and future possible methods for promoting genetic competencies considered. Phase 2 involves working with identified stakeholders within the professions and NHS to develop a strategy for future work.

(continued overpage)



Activity to date – continued

Professional groups

- The British Society of Human Genetics has drawn up learning objectives for undergraduate medical curricula.
- The Joint Committee on Medical Genetics, which operates within the Royal College of Physicians, has commissioned work to be funded by the Department of Health to devise competencies in genetics curricula for key types of specialist registrar.
- For GPs, a group under the auspices of the Royal College of General Practitioners has drafted guidance for GP Vocational Training course organisers as a prelude to getting genetic topics into the GP summative assessment and MRCGP examination.
- For nurses, a survey was published in 1999 on pre-qualification nurse education, followed by recommendations from an expert panel. The Department of Health is funding a follow-on project to map out the competencies needed for nurses in selected settings and levels.

NHS Genetics Education and Development Centre

- 4.7** Education providers, the regulatory bodies for the professions and other key organisations such as the Sector Skills Council for Health will need to consider how best to ensure genetic skills and learning needs are reflected, integrated and delivered within education curricula and training programmes for all healthcare professionals, not only for undergraduates but also for existing staff.
- 4.8** This is a major agenda. It would be unreasonable to expect existing stakeholders to take this work forward without support. **The Government is committed to ensuring that all NHS healthcare staff receive appropriate initial training and continuing professional education in genetics. We will set up an NHS Genetics Education and Development Centre which will act as a catalyst and help drive and co-ordinate activity.** The Centre will seek to work with all those bodies and organisations



responsible for determining learning needs and providing education and training to meet them. It will be affiliated to the NHSU, the developing university for the NHS, to help spread learning opportunities throughout the NHS.

NHS Genetics Education and Development Centre: Remit

The Centre will liaise with professional and academic bodies, and agencies and providers of learning in order to promote incorporation of genetics into education and training programmes for new and existing staff. It will provide assistance in:

- identifying learning needs and skill requirements for different staff groups
- developing skills and competency frameworks
- developing curricula.

It will also:

- produce learning support materials
- run seminars and workshops
- help raise the profile of genetics.

4.9 The Department of Health will invite bids from universities and other organisations with a track record in education and training to run the centre. The Centre will be quite small and staffed by a mix of specialists from education and clinical practice. It will draw on the outputs from the Public Health Genetics Unit project (see box on page 48) and work with the stakeholders for each staff group to identify what their needs are and help them meet them.

4.10 As well as targeting undergraduate and pre-specialisation training, **an early priority for the Centre will be to ensure that all GPs have access to appropriate genetics education and training when they need it to support effective practice.**



- 4.11** The Centre will not normally provide training courses directly, but it may provide some “training the trainer” programmes for teachers of genetics. It will also provide other support (such as promotion of good practice, running seminars and workshops and providing ad hoc expertise) and help raise the general profile of genetics in healthcare for example by hosting conferences and other key events.

Visiting fellowships in genetics

- 4.12** For the NHS to maintain a world leading position in the provision of genetics based healthcare, it will be essential to keep abreast of developments in genetics taking place in other countries. To maximise the inflow of genetics knowledge and practice from abroad into the NHS, **the Government will set up a scheme to enable NHS clinicians and scientists to travel abroad to acquire new genetics knowledge and expertise. We will make available a Genetics Visiting Fellowships Fund of £1.25 million over the next three years to support a number of travelling bursaries.**
- 4.13** In order to ensure that as many people as possible can benefit, the bursaries will be for a maximum period of three months. We will invite people to submit bids for funds to cover their travel and subsistence costs as well as the cost of backfilling their post where necessary. They will need to demonstrate the value of their proposed project and how they will ensure on their return that others will benefit from what they have learned.
- 4.14** In some cases it may be more cost effective to invite international experts to come to Britain to share their knowledge and expertise with the host organisation. The Genetics Visiting Fellowships Fund will be open to bids for this purpose as well.

Supporting evidence based care

- 4.15** New technologies and interventions need to have proven clinical utility if they are to be used routinely in healthcare. It will be important to show, for example, that new facilities for genetic testing will genuinely add value to clinical care. Those involved in medicines management will have to judge whether a particular pharmacogenetic test is or is not important for the safe use of medicines.



- 4.16** There is now a comprehensive range of tools and systems in the NHS that can help with these challenges. The Government is committed to ensuring that these mechanisms are deployed to support the spread of genetic based healthcare in the NHS:
- the National Horizon Scanning Centre is responsible for providing advance notice of significant new and emerging health technologies to the UK's Departments of Health. **The Centre will be including genetics in its regular programme of work.** This will ensure that awareness of key developments in genetics will be raised at a strategic level, including the prioritisation processes for the National Institute for Clinical Excellence
 - the National Health Technology Assessment programme commissions research information on the costs, effectiveness and broader impact of interventions. **Some developments in genetics may be important enough to include in the national programme. The National Genetics Reference Laboratories will also undertake health technology assessments**
 - as evidence emerges, **genetic knowledge and technologies will need to inform the development of national service frameworks.** Clinical standards developed as part of National Service Frameworks can help reduce unacceptable variations across the country and will be monitored by the Commission for Healthcare Audit and Inspection
 - clinical guidelines can make decisions about patient care easier and ensure patients receive the most effective care. The National Institute for Clinical Excellence (NICE) produces authoritative guidance for the NHS in England and Wales on the clinical and cost effectiveness of healthcare interventions and on the treatment of clinical conditions. Through its guidance, NICE can promote the faster uptake of new technologies. Healthcare professionals are expected to take NICE guidance fully into account when exercising their clinical judgement. NICE is already producing clinical guidelines in familial breast cancer and has produced appraisals for two medicinal products, trastuzumab (Herceptin) and imatinib (Glivec), that require analysis of genetic features of tumour cells before they are used. **Drawing on its advisory bodies and expert advice, the Department of Health will feed new developments in genetics, including pharmacogenetics, into the prioritisation process for NICE's work programme as the evidence base develops**



- in the absence of NICE guidelines or before an adequate evidence base has emerged, authoritative and credible advice from recognised experts can often be useful in sharing best practice.

4.17 These mechanisms – enabling good planning and forecasting, evaluation of new interventions and technologies, guidelines and standards, clinical governance arrangements, inspection and performance monitoring – will help ensure that new genetic technologies are implemented in a safe, systematic and evidence-based way.

Investing in information systems

4.18 The pace of scientific and clinical discovery in genetics means it is difficult for individual healthcare professionals to keep abreast of the wide range of knowledge and apply it for maximum patient benefit. Information to support clinical decision making will become increasingly valuable to healthcare staff. Information systems will also need to cope with new demands around recording, storing and retrieving genetic information.

4.19 The effective utilisation of genetic knowledge will receive a major boost through the *Information for Health*, the NHS Information Strategy. Substantial investment is being made available to upgrade NHS systems. Work is in progress to:

- connect all NHS clinical and management staff to the NHS network to allow access to sources of clinical information and decision support. This has already been achieved for 98% of GPs and 97% of hospital consultants
- modernise and co-ordinate the electronic storage and handling of patients' clinical records through the Integrated Care Records Service (ICRS) – the standard patient record, one per patient, which will hold all health and social care data
- ensure the confidentiality of patient information and consent to the use of information
- define national standards and specifications for the ICRS, and improve procurement.



4.20 Security and confidentiality of patient data is central to the operation of the NHS. The Department of Health is taking steps to ensure that these safeguards, and the technologies that underpin them, are robust. Work is also in progress on systems to authenticate user access and detect misuse, so that the risk of a security or confidentiality breach is minimised. A revised Code of Practice on Patient Confidentiality for the NHS is being produced to take account of the greater potential for sharing electronic as opposed to paper records. This will form part of a wider Information Governance toolkit, also in preparation, which will guide how personal health information is shared in the NHS.

Decision support systems

4.21 **The Department of Health will encourage the inclusion of genetic knowledge in decision support systems as they are specified and developed in the future.**

These could be used to:

- look for genetic features in disease
- prompt for screening or other preventive opportunities
- take pharmacogenetic factors into account in prescribing
- recall patients, perhaps years later, for whom earlier testing has identified an increased risk of chronic disease.

4.22 Computers can also help clinicians calculate genetic risk. Software is already available to help map out family history and calculate the chances of a patient inheriting a single gene disorder or a familial form of cancer. But chip technology will mean that the results of a whole range of tests can be brought together into a single assessment of risk, and this will be a complex calculation. Suitable IT packages could also be used to help explain risk to patients.

4.23 **Work is now underway to develop a genetics portal within the National Electronic Library for Health.** The Library is a new digital on-line library facility, provided by the NHS Information Authority (NHSIA). It will provide GPs and other health care professionals with access to reference works, directories, relevant websites, guidelines and other useful sources of information. It will also be available to the public through NHS Direct Online.



- 4.24** In the early years of developing this genetics website there will be a shortage of readily available material on genetics relevant to the needs of generalists and non-genetics specialists to which the site can link. **We will fund, through the NHSIA, the production of specially written material to fill this gap. This will be constantly updated to reflect the latest research evidence and will be easily and quickly available to assist GPs, nurses and other healthcare staff in consultations with their patients and in clinical decision making.**

Information for patients

- 4.25** NHS Direct is a world leading source of information and advice for patients, available both by telephone and via the internet, through NHS Direct Online. NHS Direct Online already includes information on genetic conditions and how some common conditions run in families. **The Department of Health will ensure that NHS Direct keeps abreast of developments in genetics so that patients and the public will always be able to access up-to-the-minute information about genetic influences on health and what these might mean for them and their families.**

Recording information

- 4.26** In future, genetic features (such as family history, inheritance patterns, test orders and results, diagnoses detailing genetic sub-types of disease, estimates of future risk) will need to be recorded in patients' electronic records to inform subsequent treatment decisions. To make this possible, **the Department of Health will 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.**
- 4.27** The NHS uses a structured system of clinical terms to allow healthcare professionals to record details about patients in a consistent clinical language, and to allow computers to capture and retrieve this information in a systematic way. From 2003, the standard set of clinical terms for use in the NHS will be SNOMED-CT. The Department of Health is already working with the NHS Information Authority and current and future users of genetic terminology to ensure that SNOMED-CT will cover the important genetic concepts, such as modes of inheritance and levels of risk.



Supporting commissioners

4.28 Genetics has implications for healthcare delivery across the entire medical spectrum. It presents particular challenges for commissioners. They will need to keep pace with new developments and new evidence, assess their likely impact and utility and anticipate the cost implications.

4.29 A further challenge for commissioners is to support the introduction of genetic applications at an appropriate population level. Primary Care Trusts (PCTs) now have responsibility for commissioning nearly all NHS services. Genetics is one of the specialised services that has been commissioned by the Regional Specialised Commissioning Groups (RSCGs) up until now. A review, including a consultation exercise, of commissioning arrangements for specialised services was carried out in 2002. Guidance was issued in March 2003 which emphasises that building on the work of the RSCGs, PCTs are expected to develop collaborative commissioning arrangements for specialised services.

4.30 New models of collaborative commissioning will be a feature of the UK Genetic Testing Network. The Network brings together the commissioners and genetics laboratory heads from around the UK. Led by the London RSCG, the network provides a national focus for work on:

- stability of service provision, particularly for the rare disorders
- equity of access for patients and their families via their nearest genetics centre which will act as the gateway to the network
- collaborative planning for the managed introduction of new developments showing clinical utility.

4.31 As well as supporting the commissioning of specialised genetic services, PCTs will also need to manage demand locally. As public and professional awareness increases, making sure referrals are appropriate will become increasingly important if specialised genetics services are not to be overwhelmed. To cope with the anticipated increase in demand for genetic tests and related advice, it is likely that a steady sustained increase in funding for genetic services, including testing, will be necessary. PCTs will need to factor this in to their local prioritisation in commissioning.



- 4.32** The Genetics Commissioning Advisory Group, formed in early 2001, has brought together commissioners of genetics services, genetics experts and other stakeholders to provide national strategic advice on commissioning. In collaboration with genetics professional bodies, GenCAG has produced a set of quality markers for clinical and laboratory genetics. Commissioners and service providers are now using these to benchmark services locally.
- 4.33** Recognising that PCTs now have a pivotal role in commissioning all services, GenCAG has also produced a guide for PCTs on the planning, provision and commissioning of genetics services.
- 4.34** Education and training in genetics will help prepare PCTs for their current and future role. **The Public Health Genetics Unit and the Genetic Testing Network team, with funding from the Department of Health, will identify commissioners' needs in genetics and provide a rolling programme of training to meet them. This will consist of locally tailored workshops as well as national events. Over the first year of the programme, all those involved in commissioning genetics services will be offered opportunities to participate.**

Conclusion

- 4.35** The Government will support the integration of genetics knowledge and health care applications across the NHS by:
- **setting up an NHS Genetics Education and Development Centre which will act as a catalyst to bring education and training in genetics for all NHS healthcare staff. An early priority for the Centre will be to ensure that all GPs have access to appropriate genetics education and training when they need it to support effective practice**
 - **setting up a Genetics Visiting Fellowships Fund to allow NHS clinicians and scientists to travel abroad to acquire new genetics knowledge and expertise. The Fund will also be open to bids to invite international experts to come to Britain to share their knowledge and expertise with the host organisation**



- **developing a genetics portal on the National Electronic Library for Health. Funding the production of specially written material for this website to ensure that all GPs and other healthcare staff have access to up-to-date genetics information to assist them in clinical decision making**
- **ensuring that NHS Direct keeps abreast of developments in genetics so that patients and the public will always be able to access up-to-the-minute information about genetic influences on health and what these might mean for them and their families**
- **ensuring 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**
- **supporting 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. Drawing on its advisory bodies and expert advice, the Department of Health will feed new developments in genetics, including pharmacogenetics, into the prioritisation process for NICE's work programme as the evidence base develops**
- **investing in training for commissioners. The Public Health Genetics Unit and the Genetic Testing Network team will identify commissioners' needs in genetics and provide a rolling programme of training to meet them. Over the first year of the programme, all those involved in commissioning genetics services will be offered opportunities to participate.**

5. Generating new knowledge and applications

- 5.1** Genetic research has made enormous strides since James Watson and Francis Crick determined the structure of DNA. Across the world, research into human genetics has become of prime importance and scientists are collaborating on projects of potentially huge significance to NHS patients.
- 5.2** **The Government will continue to encourage the extensive range of research initiatives underway in the UK, both in the public and private sectors. Our vision is for the UK to maintain its role at the leading edge of genetics research and development.**
- 5.3** Realising the benefits of genetic research will be a key challenge for the NHS over the coming decades. It is important to have realistic expectations about the speed with which genetic discoveries will be translated into better patient care. New therapies and diagnostics will need to be carefully examined for their clinical utility, cost effectiveness, and their ability to add value to patient care. However, the scale of the research effort in the UK and overseas should result in significant benefits for patients.
- 5.4** The UK is at the leading edge of genetic research and development. Our universities, research institutes and industries have an outstanding track record in this area and are undertaking ground-breaking research in genetic discovery and utilisation. Our biotechnology and pharmaceutical companies are making a significant investment in the research and development of genetic tests, pharmacogenetics, and gene-based therapies. A third of the Human Genome Project work was carried out in the UK, and British scientists have recently won Nobel Prizes for their outstanding work in genetics.



- 5.5** The Government, the Government-funded research councils, medical charities, universities, industry and the NHS work together to fund this world-class portfolio of UK research, covering many interacting programmes.
- 5.6** Across the EU the €17.5 billion (£12.5 billion) 6th Framework Programme for research and development (2002-2006) aims to nurture some of the best in European research, supporting a number of areas including a €2.255 billion (£1.6 billion) programme on life sciences, genomics and biotechnology. The UK Government contributed an estimated £442.6 million to the EU research and development budget in 2000/01.
- 5.7** The Government has provided an additional £1.25 billion to support UK science and technology as part of a long-term strategy for science, called *Investing in Innovation: A Strategy for Science, Engineering and Technology* published in July 2002. The strategy emphasises the importance the Government attaches to working in partnership with industry and making the UK an attractive and hospitable choice for world class companies and researchers.
- 5.8** The allocation of the Science Budget announced on 9 December 2002 included increased funding for Research Council post-genomics and proteomics research. Additional support of £110m over three years, provided in Spending Review 2000, will be extended and increased under Spending Review 2002 so that it now amounts to £246 million over 5 years. Increased capability in proteomics (understanding protein structure and function) forms a key part of the Councils' strategy to translate postgenomic knowledge into UK health benefits, including development of drugs and diagnostics. The investment will be monitored against objectives, including the international competitiveness of the science and its impact on society and the economy.
- 5.9** To complement its substantial investment in the science base, the Government has also invested £25 million in a major biotechnology programme 'Harnessing Genomics'. This is designed to help industry take up new opportunities provided by advances in biosciences, particularly genetics. The programme consists of three main elements:
- Stimulation of industrially relevant R&D and enabling technologies
 - Development of a successful UK bio-manufacturing capability



- Mentoring and incubation support to new companies and encouraging the growth of business entrepreneurial skills to enable the biotechnology sector to prosper in global markets.

Genetics knowledge parks

- 5.10 The Department of Health and the Department of Trade and Industry are investing around £15 million to support the development of five genetics knowledge parks over five years.** The genetics knowledge parks are based in London, Cambridge, the North West, Newcastle and Oxford. A sixth, the Wales Gene Park, is based in Cardiff and is being developed with funding from the Welsh Assembly Government and the DTI. They bring together a powerful combination of expertise from a range of disciplines and sectors to create an overlapping and complementary network of centres of excellence.
- 5.11** The knowledge parks will carry out research into the genetic components of major diseases, the implications of genetics for NHS services and broader ethical, social and legal issues. They will be able to work in partnership with the private sector to develop new medicines and treatments, including gene therapy, and an extended range of diagnostic tests and predictive tests. Their overall objectives will be to ensure that the NHS is better placed to exploit the findings of genetics research and to improve public understanding of genetic science. They will also provide opportunities for increasing UK industrial competitiveness.
- 5.12** The full programme for each centre is extensive and includes the following examples:
- the **Northern Genetics Knowledge Park** is searching for links between ageing and cancer. The genetic material inside cells of body tissues becomes less stable with time. The extent of this instability in normal tissues and how this changes with age may provide additional knowledge of individual susceptibility to diseases such as colorectal cancer
 - the **Oxford Genetics Knowledge Park** is investigating how certain genes influence susceptibility to cardiovascular disease in the general population. Identifying those patients with a genetic make-up that responds well to



certain medicines may improve the efficacy of existing patient treatments. Earlier screening for particular genetic features may identify individuals at risk of cardiovascular disease who could benefit from earlier intervention

- the **London Ideas Genetics Knowledge Park** is undertaking in depth family studies to gain a comprehensive understanding of genetic variants in screening, and assessing feedback on the psychological impact of genetic testing. It is also examining NHS genetic services and developing training courses and tools that will enable wider uptake and implementation of genetic services in the NHS
- the **North West Genetics Knowledge Park** is examining ways of deploying genetic health care services. This involves identifying who needs specialist genetic services and looks at equity of access for all patient groups. Genetic health care systems must also be flexible in response to the arrival of new tests and technologies and need to be responsive to informed patient choices
- the **Cambridge Genetics Knowledge Park** will be specifically examining issues of policy, ethical, social, legal and public health genetics. Intellectual property, maintaining patient confidentiality and privacy, consent, regulation and the impact of genetics on primary care will also all be explored. It will also focus on educational and training activities for health professionals, managers and entrepreneurs
- the **Wales Gene Park** will undertake research in cancer, inherited neurodegenerative diseases and psychiatric disorders. It will also look at the wider applications of genetic technologies, such as in relation to environmental genetics. Multi-disciplinary teams will be working with new small companies to help business creation. It will also investigate social issues raised by the applications of genetics, as well as models of health service delivery.

Centrally funded research

- 5.13** As well as co-funding the genetics knowledge parks, the Department of Health will sponsor new research initiatives to help convert genetic discoveries into improved patient care.



- 5.14** To provide a strategic oversight, **the Department of Health has set up a new Advisory Group for Genetics Research.** Members will offer advice on areas where additional research may be required to address the needs of the NHS. They will also monitor progress and review research funded by the Department and others to identify how it could impact on the NHS.
- 5.15** Three areas of work have already been identified by the Department of Health as research priorities: genetics based health services, pharmacogenetics and gene therapy.

Genetics based health services

- 5.16** To help the NHS incorporate genetic knowledge into patient care, the **Department of Health will invest up to £1.5 million to fund a range of research into genetics based health services.** Researchers, front line staff and users will be invited to help establish the priorities for this research.
- 5.17** These priorities might include:
- communication of genetic risk and how patients respond to individual information on risk
 - access (including ethnicity, equity and equality) issues and how beliefs affect utilisation of services with a genetic element
 - economic studies to identify the cost effectiveness of different approaches.

Pharmacogenetics

- 5.18** Research in pharmacogenetics should help make medicines usage more effective. It should allow doctors to identify patients who could suffer adverse reactions as well as those who may not respond to a particular drug at all. And it should help them tailor the dose according to a person's individual needs.
- 5.19** Pharmaceutical and biotechnology companies are investing heavily in pharmacogenetic research and the Government welcomes this far-sighted investment. However, it is mainly focused on innovative products that may not be marketed for several years.



- 5.20** To secure earlier benefits for NHS patients, **the Department of Health will invest up to £2.5 million in pharmacogenetic research on existing medicines** which patients are commonly taking now, or are likely to be taking soon. As our knowledge of drug/gene interactions expands, this money will also be available to support pilots within the NHS of pharmacogenetic testing to assist prescribing of drugs which are already on the market. We would particularly like to see greater collaboration between NHS clinicians, academia and private industry in running such pilots.
- 5.21** Given the vast number of medicines available, the Department of Health funded projects will focus on areas where pharmacogenetic influences and utility could be greatest. For example:
- studying medicines or classes of medicines which are very commonly used (such as drugs to lower blood pressure), particularly expensive or used in otherwise healthy people (such as oral contraceptives)
 - investigating serious adverse reactions which occur in response to a number of different types of medicine
 - exploring medicines whose usefulness is significantly reduced by genetic-related toxicity, so that these medicines can still be used to benefit patients who are genetically less likely to suffer toxic effects.
- 5.22** To boost this research effort still further, **the Department of Health will provide funding to set up the first university Chair in pharmacogenetics, supported by a small department of two to three full time researchers.**

Near-patient genetic testing

- 5.23** Technology is currently being developed which should allow genetic testing and DNA analysis of viruses and other microbial infections to be carried out quickly and easily by non-specialist staff in primary care or emergency settings. Such technology could be very valuable as a diagnostic tool where a rapid result is needed, for example to determine whether a patient has meningitis or not. The same technology could be used to carry out pharmacogenetic tests in future as the knowledge base to support genetic based prescribing develops. Because of the wide ranging potential of such technology, **the Department of Health would like to support the piloting of**



near-patient genetic testing in the NHS to help determine its reliability, utility and validity in clinical settings. We will make available up to £500,000 for this purpose.

Gene therapy research

- 5.24** Gene therapy offers the potential to cure or alleviate inherited conditions by introducing new genes into the body to replace or augment faulty genes. Currently, most clinical trials into gene therapy aim to treat cancer. Although there have been some noted successes in treating single gene disorders with gene therapy, this type of gene therapy research is less attractive to industry because of the small numbers of patients with a given disorder. To address this, **the Department of Health will invest up to £2.5 million to support gene therapy research into single gene disorders.** Boosting research in this area now has the potential to benefit the estimated three-quarters of a million patients in this country with single gene disorders that are currently incurable.
- 5.25** Cystic fibrosis is the most common inheritable single gene disorder in this country. There are 7,500 children and young adults with cystic fibrosis in the UK. The primary symptom of this distressing condition is that mucus continually builds up in the lungs causing breathing difficulties and respiratory infections. Patients can also suffer gastro-intestinal and fertility problems. There is no cure for cystic fibrosis although regular physiotherapy, antibiotics and enzyme supplements can slow the progression of the disease. Current life expectancy for children born with cystic fibrosis is about 30 years. In addition to the funding set out above, **the Department of Health will make available £2.5 million over 5 years to support gene therapy research for cystic fibrosis.**
- 5.26** We will also take action to remove barriers to gene therapy clinical research for NHS and public sector researchers who do not have commercial sponsorship. One major difficulty is the cost of producing vectors, which have to deliver the new genetic material to the appropriate part of the body. These substantial costs will increase further once the new clinical trials directive becomes part of UK law in May 2004. The Clinical Trials Directive (2001/20/EC) requires higher standards for the manufacturing of materials for clinical trials, including gene therapy vectors.



- 5.27** In order to ensure that NHS researchers are not disadvantaged by these new requirements **the Department of Health will invest up to £4 million to provide NHS and other public sector researchers with access to high standard gene therapy vector production facilities.** This funding should be sufficient to meet the full need for gene therapy vectors within the NHS for both research and treatment over the next five years.
- 5.28** As with any branch of medicine it is not possible to know in advance of extensive clinical experience what the full range of potential side effects of gene therapy might be. For example, some gene therapy treatments use a virus to deliver the therapeutic gene into chromosomes of the patient's cells. However the process is largely random and the virus could insert anywhere in the patient's genome, potentially leading to disruption of normal genes. **The Department of Health will fund research into the long-term safety of the use of gene therapy vectors which are designed to insert into human genetic material.** This research should be invaluable in minimising the risks and maximising the benefits of gene therapy.

Intellectual property

- 5.29** The NHS employs a large number of talented people. To ensure that innovations created by NHS staff have a better chance of being introduced into clinical practice for the benefit of NHS patients and society as a whole, new guidance on the exploitation of intellectual property has been produced. The document *The NHS as an Innovative Organisation: A Framework and Guidance on the Management of Intellectual Property in the NHS* sets out how NHS trusts and primary care trusts can manage and exploit intellectual property. To support NHS intellectual property exploitation a series of NHS technology transfer organisations or “hubs” is being created throughout the UK. Together, the new guidance and hubs should lead to more good ideas being developed by the NHS itself, with a rightful share of profits returning to the NHS to support further innovation, bringing better care to patients.
- 5.30** Patents are vital in encouraging innovation and competitiveness in the biotechnology sector, enabling companies to protect their investment in research to produce new and improved healthcare products for the future. Competition and patent law provide extensive measures to ensure that anti-competitive practices do not prevent the commercial development of ideas or research. These measures include the



Government's use of compulsory licensing, should a company seek to act in such a way as to prejudice the provision of NHS medical services, such as genetic testing services. The use of compulsory licensing is subject to reasonable remuneration to the patent holder.

- 5.31** The Government recognises that there are concerns about the impact patents on genetic sequences may have on healthcare. **To examine this issue in detail, the Department of Health has commissioned an independent study into the impact and management of intellectual property rights and genetics within the healthcare sector.** The study is proceeding in two stages. The first stage is complete and a report entitled *Intellectual property rights and genetics: A study into the impact and management of intellectual property rights within the healthcare sector* was published in September 2002. A conference to discuss the second stage of the study was held in February 2003 and the final report is due to be completed later this year.
- 5.32** The DTI has also commissioned a separate study on the impact of current UK law and practice regarding patents for genetic sequences more generally. This will be completed by the end of 2003. The study will review current UK law and practice regarding patents for human, plant, animal or micro-organism genetic sequences and the impact this has on the bioscience sector in the UK. This will include the impact on academic and public research, biotechnology and pharmaceutical companies. It will assess this impact in terms of the encouragement of research, the volume and type of research carried out, the commercialisation of innovations and the short and long term relations between stakeholders, taking into account relevant external practices. The study will also compare current UK practice, and its effects, to those of the US and other EU member states, and assess the consequences for the competitiveness of the UK as a choice for research and its exploitation.
- 5.33** These two separate and complementary reports fulfil the commitment made in the Government's response to the report on human genetic databases by the House of Lords' Select Committee on Science and Technology. The Government will consider them both carefully.



The UK Biobank project

- 5.34** The Department of Health, the Wellcome Trust and the Medical Research Council are to provide an initial £61 million to support the development of the UK Biobank project. This project aims to obtain comprehensive data on the combined effects of genotype, life style and environmental exposure to assess the risk of developing the common multi-factorial diseases of later life.
- 5.35** The UK Biobank will be a huge prospective cohort of some 500,000 men and women volunteers aged between 45 and 69 who will be recruited through general practices. It will hold data on health and lifestyle plus blood samples for biochemical and genetic analysis. There will be long-term follow up of the cohort via NHS medical records to accumulate data on health outcomes. It is anticipated that the project will begin later in 2003 and that it will run for at least 10 years. It will be the largest database of its kind in the world.
- 5.36** A company limited by guarantee will be created to take responsibility for the overall management of the project, the databases, financial control, quality control and audit of all the scientific activities. The recruitment of participants, the collection of baseline and health outcome data, and the collection of blood samples for biochemical and genetic analysis will be managed on a regional basis. A science committee will be responsible for agreeing any changes to the protocol and monitoring scientific output.
- 5.37** An independent monitoring body will also be established to ensure that samples of genetic material are taken with the fully informed consent of the participants and that procedures to protect confidentiality are strictly adhered to. This body will have the power to veto uses of the data or samples that it considers to be against the interests of the participants or likely to damage the reputation of the study.

Access to information in genetic databases

- 5.38** There have been concerns about the police or other law enforcement agencies seeking access to genetic databases as part of criminal investigations. During the planning of the UK Biobank, individuals and researchers expressed concern that



some people may be deterred from participation in health research studies if the police were able to compare DNA recovered from crime scenes with the donor samples.

- 5.39** The Human Genetics Commission has recommended that genetic databases established for health research should not be used by the police or law enforcement agencies to investigate criminal activities.
- 5.40** The Government takes these concerns seriously, whilst at the same time recognising the possibility of situations where there is a strong public interest to solve a particularly serious crime such as murder. Information in such genetic databases will not normally be accessible to the police as it is excluded material under the Police and Criminal Evidence Act 1984. Access to excluded material would have to be via a production order or warrant granted by a circuit judge under the Police and Criminal Evidence Act 1984 and the police would need to have clear grounds to apply for such an order. The Government expects that requests by the police for such access would only ever arise in the most exceptional circumstances.

Regulation of research

- 5.41** We will continue to actively encourage the proper ethical oversight of all human genetic research. Much of the UK's clinical research is conducted within the NHS. Research projects which involve NHS patients, staff or medical records must be reviewed by a research ethics committee to ensure the fundamental principles of consent, confidentiality and data security are not breached. Independent oversight by research ethics committees is also required by the main research councils and all the major research charities and industry bodies.
- 5.42** The Government also notes the HGC's recommendation that all genetic research on non-anonymised human tissue samples or bodily materials should be subject to review by an independent research ethics committee and that there should be arrangements for monitoring and compliance. We support this view and it is part of the Department of Health's framework for research governance, which is due to be implemented in all NHS care organisations by 2004.
- 5.43** The Government will continue to monitor the conduct of genetic research in the UK and will take action to introduce checks and balances where necessary.



Conclusion

5.44 The UK is at the forefront of generating new genetic knowledge and applying it in healthcare. The Government is committed to maintaining our leading role. It will bring enormous advantages to the NHS and the economy.

The Government:

- is investing around £15 million to support the development of six genetics knowledge parks over five years. The genetics knowledge parks are based in London, Cambridge, the North West, Newcastle, Oxford and Cardiff. They bring together a powerful combination of expertise from a range of disciplines and sectors to create an overlapping and complementary network of centres of excellence
- will invest up to £1.5 million to fund a range of research projects in the area of genetics based health services
- will invest up to £4 million to fund pharmacogenetic research on existing medicines for the benefit of patients
- will provide funding to set up the first university Chair in pharmacogenetics, supported by a small department of two to three full-time researchers
- when the technology becomes available, will provide up to £500,000 to support piloting of near patient genetic testing in the NHS to help evaluate its reliability, utility and validity in clinical settings
- will invest up to £3 million to support gene therapy research on single gene disorders
- in addition, will provide £2.5 million over 5 years to support gene therapy research for cystic fibrosis, the most common single gene disorder in the UK
- will invest up to £4 million to provide access to high standard facilities for gene therapy vector production for NHS and other public sector researchers



- **will publish an independent study into the impact and management of intellectual property rights within the healthcare sector**
- **will continue to monitor the conduct of genetic research in the UK and will take action to introduce checks and balances where necessary.**

6. Ensuring public confidence

6.1 Advances in genetics offer the potential to transform health care. Increasing understanding of the part genetics plays in the development of disease will lead to more precise diagnoses and new drugs and therapies. Knowledge about our individual genetic make-up will help to pinpoint the diseases to which we are most susceptible and lead to more personalised, effective treatment. Against this background of great promise, very real ethical and social concerns have to be considered.

6.2 Given the extraordinary pace at which genetic science is moving and the potential applications of some of the breakthroughs, it is not surprising that many of us view genetics with a mixture of optimism and apprehension. Some people fear that genetic technologies will be used in ethically unacceptable ways and that genetic advances will change our lives forever in ways that we may not welcome and are powerless to resist.

6.3 The Government recognises and understands these concerns and shares many of them. We want to maximise the benefits of genetic research but this will not be possible without public acceptance and public confidence. We need to be alert to the potential adverse consequences and prepared to take action where necessary.

The Government is committed to ensuring:

- **greater public understanding of genetics through transparency and openness and encouraging wider public debate**
- **that robust, timely and proportionate controls are in place**
- **that we receive expert and independent advice on genetics issues.**

Key safeguards

6.4 We have already put key safeguards in place against:

- unacceptable use of genetic tests by insurance companies



- the possibility of human reproductive cloning
- uncontrolled use of genetics in reproductive medicine
- uncontrolled development of gene therapy.

Moratorium on use of genetic tests in insurance

- 6.5** People could be discouraged from taking genetic tests if they had to declare adverse results that then made insurance unobtainable or unaffordable. There could also be financial implications for insurers if people with adverse test results subsequently took out high value policies while concealing their risk.
- 6.6** The Government made a manifesto commitment to tackle this issue and we have done so. We have negotiated a moratorium with the insurance industry that will last until 2006. Under the moratorium, insurers may not use genetic test results in setting premiums for life insurance policies up to £500,000 or for critical illness, long term care and income protection policies up to £300,000. Above these limits, insurance companies are only allowed to use results of genetic tests that have been approved by the Government's Genetics and Insurance Committee. To date, the only genetic test that has been approved by the committee is for Huntington's disease for life insurance policies over £500,000. The Committee also has a vital role in monitoring compliance with the moratorium and for investigating any complaints that cannot be resolved by the insurance company or the Association of British Insurers.
- 6.7** Any failure of the agreement will lead to reconsideration of the need for statutory control. If there is evidence of non-compliance with the moratorium by the insurance industry, then the Government is prepared to enforce it through legislation. We are also committed to working with patient groups and with the industry to ensure a longer-term solution.

Banning human cloning

- 6.8** A few maverick scientists are reported to be seeking to use advances in genetic science to try to clone people. The Government shares the public's repugnance that such an experiment could be attempted. We made a manifesto commitment to prevent this from happening in the UK and we have acted swiftly to deliver that



commitment by passing the Human Reproductive Cloning Act at the end of 2001. This explicitly bans any attempt to create a cloned human baby in the UK. Since then we have been actively supporting the United Nations' attempt to introduce an international ban on human reproductive cloning and we will continue to do so.

Regulating the use of new reproductive techniques

- 6.9** Genetic advances can offer real benefits in the field of reproduction and fertility treatment. A technique known as preimplantation genetic diagnosis (PGD) allows embryos created through *in vitro* fertilisation (IVF) to be tested for a specific genetic condition before being placed in the woman's womb. In this way, couples at risk of having a child with a serious inherited disorder can help ensure that their baby is free of the condition. This technique is still developing and success rates are not yet high, but for some couples this is a preferable alternative to prenatal testing and possible abortion (subject to the provisions of the Abortion Act). The range of genetic conditions for which PGD is possible is continually increasing and examples include Duchennes muscular dystrophy, cystic fibrosis and beta thalassaemia.
- 6.10** It is essential that all new genetic techniques in assisted reproduction are properly regulated to ensure that they are ethically acceptable and safe. The Human Fertilisation and Embryology Authority is the independent statutory body, established in 1991, to regulate the provision of assisted conception services and embryo research in the UK. Any new genetic techniques for screening, testing or manipulating embryos can only be used if approved by the Authority. All clinics wishing to offer PGD for a particular genetic condition must first obtain specific licenses from the HFEA. The HFEA was the first body of its kind in the world and it now provides an invaluable safeguard in this area by ensuring that genetic advances in reproduction are used only for serious medical purposes and only in a responsible and appropriate way.

Regulating gene therapy

- 6.11** Gene therapy offers enormous potential to cure or alleviate inherited conditions and common diseases such as cancer, and the Government is committed to harnessing these benefits including through the promotion of research, as set out in chapter 5.



6.12 But there are also legitimate concerns about its application and safety. The Gene Therapy Advisory Committee, an internationally respected independent body, addresses these issues. All gene therapy trials in the United Kingdom must first receive prior approval by the Committee, which considers and advises on the acceptability of proposals on ethical grounds, taking account of the scientific merits of the proposals, the potential benefits and the risks. GTAC not only monitors trials whilst in progress – for example requiring rapid notification of any adverse events – but now has a unique programme to monitor the general health and wellbeing of all UK gene therapy patients throughout their lifetime. As with all medicines, gene therapy products also have to be approved for use by the Medicines and Healthcare Products Regulatory Agency.

Increasing public awareness and understanding

6.13 The Government has intensified its efforts to engage the public in the fast-moving world of science and technology. The Prime Minister emphasised in his speech at the Royal Society that “we need better, stronger, clearer ways of science and people communicating. The dangers are in ignorance of each others’ point of view; the solution is in understanding them”.

6.14 Openness and transparency in policy making on science is an essential first step. A number of influential reports have helped to shape the Government’s approach. Examples are Lord Phillips’ Inquiry into BSE, the House of Lords Science and Technology Committee report on Science and Society, both in 2000, and the House of Commons Science and Technology Committee report on the scientific advisory committee system in 2001. There are now clear guidelines on how the Government receives scientific advice and takes account of uncertainties and the plurality of opinions. In addition, under the Office of Science and Technology Code of Practice, scientific advisory committees are required to make information available to the public and take account of public views. These important principles are drawn together in a recent Cabinet Office report *‘Risk: Improving Government’s capability to handle risk and uncertainty’* published in November 2002.

6.15 The scientific community is responding to the need for wider public involvement. The research councils, research charities and bodies such as the Royal Society pay particular attention to their role in increasing public awareness and engagement. The Royal Society launched its Science in Society programme last year to promote



dialogue with the public about science. As part of this, their recent National Forum for Science brought together members of the public, scientists and other stakeholders to consider the issue of genetic testing. Another example is the work of the Science Media Centre, based at the Royal Institution. This was launched to improve science communication and provide a resource for journalists. The Government warmly endorses these developments.

- 6.16** The broadcast and print media also have an influential role. The BBC (in collaboration with the Wellcome Trust and others) has made human genetics the subject of a number of responsible and accessible television and radio programmes and has a large amount of educational material on its website.
- 6.17** The Government has made a major investment in establishing six new genetics knowledge parks, described in chapter five. As part of their core remit, the knowledge parks will encourage local debate about the ethical and social implications of advances in human genetics.
- 6.18** Many other bodies in the public, private and voluntary sector play an important part in increasing public awareness and knowledge, and providing a full spectrum of independent views.
- 6.19** The Progress Educational Trust, a voluntary organisation part funded by the Department of Health is dedicated to improving people's understanding of genetics. The Trust organises conferences and seminars and works with schools and universities to provide genetic education for young people. It also publishes a range of briefings including a weekly internet newsletter called Bionews which is available free to anyone who requests it.
- 6.20** **The Department of Health will provide an additional £200,000 of funding to the Progress Educational Trust over the next three years to build on the valuable work they have done to date.** This additional funding will allow the Trust to develop a dedicated Bionews website, run an extensive national programme of public debates on topical issues in genetics and produce an updated *'Guide to Genetics'* booklet which will provide an accessible introduction to genetics for the lay reader.



6.21 The Government is also providing around £1,000,000 to support a programme of events and initiatives in 2003 to celebrate the 50th anniversary of the discovery of the structure of DNA. Other organisations such as the British Council are also playing a major part. A key aim of this programme will be to increase people's awareness and understanding of genetics and the potential it offers.

Anticipating and addressing issues of public concern

6.22 We have already put key safeguards in place, and we are committed to encouraging greater public awareness and understanding of human genetics. These have been prerequisites to ensuring acceptance of the application of new genetic knowledge. **The Government is also committed to ensuring that its regulatory framework around genetics and health anticipates and reflects public concerns.**

6.23 The Human Genetics Commission (HGC) has a critical role to play here. The Government established the HGC in 1999 as an independent UK wide body to provide expert advice on human genetics and the social and ethical issues that arise. In conducting its work, the HGC has been a model of openness and transparency. It has sought innovative ways of engaging the general public and ensuring that people with genetic conditions are represented. In 2001 it set up a consultative panel consisting of around 100 individuals with direct experience of a genetic disorder, either personally or in their family. This panel will be a valuable resource for the HGC in developing their thinking.

6.24 In 2001 the HGC published a major survey into how the public feel about different aspects of human genetics. This showed that most people broadly supported the use of genetic information in healthcare and criminal investigations. But in line with other findings, a significant proportion were concerned about the wider uses of genetic information.

6.25 As a result, the HGC gave lengthy consideration to the legal and ethical issues surrounding the use of personal genetic information in its report *Inside Information: Balancing interests in the use of personal genetic data*. The conclusions that the HGC reached and the ethical principles it laid out (page 86) have informed the Government's thinking in this area. The HGC concludes that the sensitivity of genetic information depends entirely on the nature of that information and the context in which it is considered. For example, in a strict sense, a person's blood



group is genetic information, but there are very good reasons why people want that information to be widely known by healthcare staff.

- 6.26** We warmly welcome HGC's clearly-argued overarching principles, especially the balancing of respect for individual rights (for privacy, confidentiality and non-discrimination) with the need for "genetic solidarity and altruism" so that genetic knowledge can be shared to help others and society. These two principles form a valuable basis for developing future Government policy on genetics.

Protection of genetic information

- 6.27** One example of the delicate balance between individual and wider interests considered by the HGC is the practice of recording information about family members in medical records. This is especially true of records held by clinical genetics centres that rely on carefully recorded family histories. Under the Data Protection Act, a doctor or counsellor is required to tell the relatives that information about them is recorded in the patient's medical records. Not only is this potentially very bureaucratic, it could also reduce the amount of clinically useful information that doctors feel they are allowed to record. The HGC also said it was possible in some cases that a relative may wish to stop the information about them being recorded. The patient's interests would then have to be balanced against those of the relative.
- 6.28** The Government has taken note of the comments made by the HGC and of other concerns about the application of the Data Protection Act to medical record-keeping. We established the Health Records and Data Protection Review Group to examine the matter in detail. They are due to report in 2003 and we look forward to receiving their recommendations.
- 6.29** One of the major recommendations of the HGC report was to create a new criminal offence of testing an individual's DNA without their knowledge or consent for non-medical purposes. HGC made this recommendation to prevent people trying to obtain DNA covertly (for example by taking discarded hair or dental floss from a dustbin) and testing it without the individual's consent. The intention behind such non-consensual testing would be to acquire sensitive information which could then be used in undesirable ways, for example to blackmail the person or to sell a story to the media.
- 6.30** The Government has consulted on this proposed new offence as part of "*Human Bodies, Human Choices*", the review of the law concerning the removal, retention and



use of human tissue and organs. We have taken note of the responses that we have received. In framing any new offence it will be essential that:

- it does not interfere with the use of genetic material by the police or the courts
- it must not result in doctors or researchers being unsure when and whether they can order new genetic tests without returning to the patient for consent
- the proper lawful access to private paternity testing is not affected.

6.31 With these provisos in mind, **we will introduce a new offence of testing an individual's DNA without their knowledge or consent** by means of a new Bill that will regulate the removal, storage and use of human organs and tissues. The new offence will not cover medical situations where consent is impossible to obtain, or use by the police and courts. We believe that this new offence will provide an effective means of ensuring the privacy of individuals and provide a proper deterrent against the wrongful testing of genetic material. This will be a UK-wide offence.

Preventing unfair discrimination

6.32 Concerns have been expressed that employers could use personal genetic information to make decisions about an individual's job application, promotion prospects or pension. Or that genetic testing could potentially be used in future as a way of selecting people in education or other areas.

6.33 A core ethical principle in the HGC report is that no one should be unfairly discriminated against on the basis of his or her genetic characteristics. The Government wholeheartedly endorses this principle. It has been explicitly stated in a number of important international treaties (such as the Council of Europe's Convention on Human Rights and Biomedicine) and declarations (e.g. the UNESCO Universal Declaration on the Human Genome and Human Rights) that the UK has supported.

6.34 The Government asked the Disability Rights Commission and the HGC to consider whether the Disability Discrimination Act should be amended to include people with a pre-symptomatic genetic condition. Whilst both bodies felt that there was a very real danger of unfair discrimination, they differed in their view about the role of the Act in preventing this. However, both bodies agreed that there should be a long-



term consideration of the possible need for specific legislation to prevent unfair discrimination of people on the grounds of their genetic characteristics.

- 6.35** We agree with the HGC that this is an extremely complex matter and will become increasingly relevant with advances in the use of predictive genetic information by the NHS and privately for occupational health purposes. Although this is not yet a widespread problem, **the Government accepts the HGC’s recommendation. We will consider the evidence for unfair discrimination on the basis of a person’s genetic characteristics and the appropriate means of addressing any concerns in this area.** We will also ask the Human Genetics Commission, with others such as the Disability Rights Commission, to continue to monitor developments.

Consent to genetic testing

- 6.36** Where a person is able to give their consent to a genetic test, that consent must be obtained before the test is carried out. The consent must be given voluntarily and the person must have received appropriate information. The legal provisions that cover the testing of children and young people, and adults unable to consent, are more complex. Guidance on those provisions is provided in the Department of Health’s Reference Guide to Consent for Examination or Treatment (2001).
- 6.37** The question of what constitutes “appropriate information” is particularly complex in genetics. Essentially, the nature of the information provided should be appropriate to the nature of the test. The HGC makes a distinction between tests that are of low “informational impact” or significance for an individual, such as tumour genotyping, and tests of high significance which may have a major impact on the patient and possibly the patient’s blood relatives. Such tests may have wider implications which could affect the patient’s financial and employment prospects as well as their social relationships and psychological wellbeing. An example of such a test would be presymptomatic testing for Huntington’s Disease.
- 6.38** The HGC recommends that for low impact genetic tests, it is not necessary to provide information on the genetic dimension of the test to the patient as long as they understand the purpose of the test. The information necessary to obtain consent to high impact genetic tests has been extensively considered by genetic counsellors and clinical geneticists and is helpfully summarised by the HGC in its report *Inside Information*.



- 6.39** As the genetic basis of more common diseases is recognised, other clinical specialists and general practitioners will increasingly become involved in requesting genetic tests. It is essential that they should be aware of the need for appropriate information to be provided to the patient.
- 6.40** Consent to a genetic test covers both the procedure for obtaining the material (such as a blood specimen) and the testing process itself. Consent for a high impact genetic test should be given in writing. But this is not necessary for tests of low impact. It is the responsibility of the clinician requesting the test to obtain the patient's consent before referring it to a laboratory. Laboratory staff should not be expected to double-check that this has been done.
- 6.41** Where consent has been given for a test for clinical purposes, it is the Government's view that it is acceptable to use any leftover material for the purposes of quality assurance and audit without requiring separate consent for those purposes, so long as patients are informed in general terms about such uses (for example through the use of notices or leaflets). Quality assurance and audit are essential in ensuring that the high quality of care that patients have the right to expect is delivered.
- 6.42** However, different procedures may be necessary if it is proposed to use tissue for other purposes, such as research. The Department of Health has recently consulted on a draft interim statement on the ethically acceptable use of human organs and tissue under the present law, pending new legislation on human organs and tissue. This interim statement was published in April 2003.

Direct to the public genetic testing

- 6.43** Alongside NHS genetic testing, "over the counter" tests are increasingly being offered to people directly by private companies and via the Internet. The issues surrounding commercial genetic testing services were considered by the Advisory Committee on Genetic Testing in 1997 and a voluntary code of practice has been in place since then. However, the type of genetic testing available is beginning to expand and is no longer confined to a limited range of tests that can predict or diagnose very serious conditions, or the risk of passing on such conditions to a child. Some genetic tests are claimed to be predictive of reactions to chemicals, drugs, foods or environmental factors and to offer the possibility to improve one's health by avoiding these factors, eating certain foods and so on. There is also a growing industry supplying DNA tests to establish family relationships or historical roots.



- 6.44** The Government supports the view that individuals who want to take a more direct responsibility for their own health should not face arbitrary barriers which restrict their access to services unnecessarily. It is arguable that genetic tests are not intrinsically different from other health test kits that are freely available over the counter such as cholesterol, diabetes and pregnancy testing kits. There are a range of general consumer protection laws that govern the claims that can be made about a product and how it is marketed. A further important safeguard in the UK is the availability of a large number of different genetic tests via the normal GP referral arrangements to a consultant or clinical genetics centre. Such tests will continue to be free at the point of delivery and to include proper pre- and post-test counselling and support.
- 6.45** Nevertheless, there are potential problems with unfettered access to genetic tests. Not all tests are simple to interpret without access to other clinical information. In some cases people need considerable counselling before and after a test to understand the limitations of a test as well as the implications for them and their family. As well as the ethical difficulties of not giving proper information before a test, the practical consequences of poor support could have an impact on the NHS, particularly GPs and geneticists who may be called upon to give people further advice and counselling. There is also a risk that the test may have uncovered the genetic basis of a serious medical condition that had not been specifically tested for. Even those genetic tests that do not result in predictive health information may be used to encourage people to take additional dietary supplements or to make other life-style changes without there being good justification or evidence.
- 6.46** In view of these concerns, the Government asked the HGC to carry out a full review of the issues surrounding the sale of genetic tests direct to the public and to consider whether further regulation may be necessary. The HGC's report *Genes Direct* was published in April 2003. We will consider its conclusions and recommendations carefully and respond in due course.

Paternal testing

- 6.47** One type of genetic testing which is increasingly being advertised direct to the public is DNA testing to establish whether a man is the father of a child. In many cases there will be a potential for disputes about who is eligible to consent to the test on behalf of the child. The Government has made changes to the arrangements for parental



responsibility in the Adoption and Children Act 2002. As a result, more men will in future be able to acquire parental responsibility. This will give them the authority to have their children tested to establish paternity.

- 6.48** In our view this makes it all the more important to ensure that there are proper safeguards in place to ensure that third parties – such as other relatives or those with no family connections – do not intrude on the privacy of families and children. At present this area is covered by a voluntary ‘*Code of Practice and Guidance on Genetic Paternity Testing Services*’ published by the UK Health Departments in 2001. However, we recognise that this has a limited scope and does not contain compliance monitoring or enforcement mechanisms. We therefore look forward to further advice from HGC on the particular aspects of consent and confidentiality that are raised by DNA paternity testing and an appropriate regulatory framework.

Genetics and behaviour

- 6.49** Until now, most human genetics research has focused on the link between our genes and our health. However, some scientists believe that human behaviour as well as human health may be influenced by the genes we inherit. Researchers in the field of behavioural genetics are attempting to locate specific genes, or groups of genes, associated with behavioural traits and characteristics such as intelligence and antisocial behaviour. This is a highly complex area, not least because of the difficulty of separating out the other factors which clearly influence our behaviour such as our upbringing and our environment.
- 6.50** It is important to emphasise that so far, no genetic variant has been shown conclusively to influence behavioural or personality traits and there are currently no practical applications of research into behavioural genetics. It is also important not to overestimate the predictive power of genes.
- 6.51** The Government recognises that this is a highly controversial area of research that will cause many people deep unease. We need to start considering now the ethical, legal and social issues that it raises. In October 2002, the Nuffield Council on Bioethics published the first major report to address these issues: *Genetics and Human Behaviour: the Ethical Context*.



- 6.52** The report begins by considering whether research into behavioural genetics should be allowed at all because of the potential for abuse. However it concludes that this type of research has the potential to advance our understanding of human behaviour and that it can therefore be justified, although it will be important to create safeguards to protect against its misuse. We agree with this view.
- 6.53** The report also raises the possibility that genetic information about behaviour might be used in the contexts of education, employment, insurance and the criminal justice system. The Government has noted these concerns and will ensure that they are taken into account in considering the need for legislation to protect people against unfair genetic discrimination. In the meantime, the Government will ask HGC to monitor research into behavioural genetics and its possible applications and to advise Government of any further action that may be needed.

Conclusion

- 6.54** Realising the potential benefits of genetics requires public acceptance and confidence. **The Government fully recognises that developments in genetics will present new ethical challenges. The Government:**
- **has developed a robust and proportionate regulatory framework around genetics and health. The Government has already introduced key safeguards – the moratorium on unacceptable use of genetic tests by insurers, banning human reproductive cloning, and regulating the use of genetics in reproductive medicine and clinical trials of gene therapy**
 - **is committed to ensuring openness and transparency in genetic policy making**
 - **will continue to promote public understanding of genetics, including through providing additional financial support to the Progress Educational Trust**
 - **is providing around £1,000,000 to support a programme of events and initiatives in 2003 to celebrate the 50th anniversary of the discovery of the structure of DNA. A key aim will be to increase people's awareness and understanding of genetics**



- **will ensure that the regulatory framework anticipates and continues to address public concerns**
- **has established the independent Human Genetics Commission to provide expert advice on developments in human genetics and the social and ethical issues raised. The Government welcomes their report *Inside Information* on the uses of personal genetic information, and in particular the ethical principles it sets out**
- **will introduce legislation making it an offence to test a person's DNA without their knowledge or consent except as part of their medical treatment where consent is impossible to obtain, or use by the police or courts**
- **will consider the evidence for unfair discrimination against people on the basis of their genetic characteristics and the appropriate means of addressing any concerns in this area.**



HGC ethical principles (from Chapter 2 of *Inside Information*)

Genetic knowledge may bring people into a special moral relationship with one another. We propose the following concept of *genetic solidarity and altruism*, which promotes the common good:

We all share the same basic human genome, although there are individual variations which distinguish us from other people. Most of our genetic characteristics will be present in others. This sharing of our genetic constitution not only gives rise to opportunities to help others but it also highlights our common interest in the fruits of medically-based genetic research.

We share the assumption of our society that individual persons have the highest moral importance or value. We therefore express the following key principle of *respect for persons*:

Respect for persons affirms the equal value, dignity and moral rights of each individual. Each individual is entitled to lead a life in which genetic characteristics will not be the basis of unjust discrimination or unfair or inhuman treatment.

We believe that a number of secondary principles may be derived from this overarching principle, taking account of the possible claims of genetic solidarity and altruism:

The principle of privacy

Every person is entitled to privacy. In the absence of justification based on overwhelming moral considerations, a person should generally not be obliged to disclose information about his or her genetic characteristics.

The principle of consent

Private genetic information about a person should generally not be obtained, held or communicated without that person's free and informed consent.

The principle of confidentiality

Private personal genetic information should generally be treated as being of a confidential nature and should not be communicated to others without consent except for the weightiest of reasons.

The principle of non-discrimination

No person shall be unfairly discriminated against on the basis of his or her genetic characteristics.

Annex A: Genetics services in Scotland, Wales and Northern Ireland

Scotland

Under the terms of the Scotland Act 1998, human genetics is a matter that is reserved to the UK Parliament. Legislative competence rests with Westminster, national policy is developed by the UK Government, in consultation with the devolved administrations among others, and the implementation of policy will be with each of the administrations depending on the means required for implementation.

There is strong research and development activity in Scottish universities and in the biotechnology industry in Scotland. The provision of NHS genetics services in Scotland is comprehensive and high quality. The four Regional Genetic Centres (Aberdeen, Dundee, Edinburgh and Glasgow) provide clinical and laboratory genetic services both within their own areas, and through their outreach clinic network, to all parts of Scotland and the Islands. Within this framework they also deliver a co-ordinated cancer genetics service. The Molecular Genetics Laboratories within the four centres are well equipped and have for some years now worked as a formal Consortium, which is co-ordinated, commissioned and funded at national level for NHS Scotland. The work of the Consortium is kept under regular review in order to ensure that the service provided reflects developments in the field, and thus recently some aspects of the work of cytogenetic laboratories (those which involve DNA based molecular tests) have been added to the Consortium. The Consortium arrangements mean that there is equity of access to all available molecular genetic tests for any Scottish patients. In addition, Scottish patients have access to any tests not available within Scotland, as national funding is available to purchase such tests from providing laboratories either elsewhere in the UK or abroad.

The implications of this White Paper for the development of services in Scotland is a matter for the Scottish Executive.



Wales

Wales has an international reputation for the quality and delivery of its Medical Genetics Service, which has led the development of cancer genetics and neuro-genetics services. It has also had a key UK role in developing standards of clinical governance and has pioneered equity in access and delivery.

The existing foundation of genetic services in Wales is strong. To ensure this can be successfully built upon a comprehensive Review has been undertaken to identify the issues arising from the world-wide rapid scientific developments being faced by Medical Genetics Services and what this means to the people of Wales. The Review identifies what is needed to provide the best possible service for people in Wales and to keep Wales in the forefront of developments.

The Welsh Assembly Government has provided an additional recurring investment of £1.5 million to develop the future of Medical Genetics Services in Wales and will consider the implications of this White Paper alongside the outcome of the Review in taking that development forward.

Northern Ireland

Clinical genetics services are provided on a regional basis for the population of Northern Ireland, with outreach clinics offered throughout the region. The Department of Health, Social Services, and Public Safety is currently conducting a review of clinical genetics services in Northern Ireland. The Review takes account of national and international developments in genetics, the increasing workload in clinical genetics, and the challenges of providing timely access to services. The aim of the Review is to shape the strategic direction for the service over the next 5–10 years to ensure high quality services which are patient-centred, co-ordinated, delivered in a timely manner and ensure equity of access.

The Review, led by the Chief Medical Officer, commenced in October and is expected to report by summer 2003. It specifically addresses workforce issues, access to services, information requirements, training for health professionals and facilities.

It is expected that implementation of the Review's recommendations will make a significant contribution to improving the capacity of clinical genetics and to reducing waiting times for patients. The implications of the White Paper will be considered in conjunction with the outcome of the Review.

Annex B: Advisory Panel

This document was informed by the work of an Advisory Panel. The Government is grateful to the members of the Advisory Panel for their contribution.

Membership

Lord Turnberg	Former President of the Royal College of Physicians (Chair)
Prof Martin Bobrow	Professor of Medical Genetics, Cambridge
Prof John Burn	Professor of Clinical Genetics, Northern Genetics Service and Director, Newcastle Genetics Knowledge Park
Dr Paul Debenham	Head of Life Sciences and Forensics, LGC
Joanie Dimavicius	Former Chair of the Genetic Interest Group
Prof Peter Farndon	Professor of Clinical Genetics, Birmingham and former chair of the Joint Committee on Medical Genetics
Penny Guilbert	Nurse/Counsellor, Nottingham City Hospital
Prof Sir David King	Government Chief Scientific Adviser, Office of Science and Technology
Crispin Kirkham	Former Chief Executive, Bioindustry Association
Delyth Morgan	Chief Executive, Breakthrough Breast Cancer
Baroness Onora O'Neill	Former Chair of the Nuffield Council on Bioethics and of the Human Genetics Advisory Commission; Principal of Newnham College, Cambridge
Dr Allen D Roses	Senior Vice-President, Genetics Research, GlaxoSmithKline; nominated by the Association of the British Pharmaceutical Industry
Dr Nigel Starey	GP and Director of the Centre for Primary Care, University of Derby
Dr Ron Zimmern	Director, Public Health Genetics Unit and Director, Cambridge Genetics Knowledge Park



Glossary

Adult-onset disorder: A **genetic condition** that shows symptoms only in later (adult) life.

Bioethics: The study of ethical problems arising in the field of biology and medicine including those arising from biological research and its applications in such medical treatments as organ transplantation and artificial insemination. The Department of Health has responsibility for bioethics within Government.

Bioinformatics: The application of computer and statistical techniques to the management of biological information such as **DNA** sequences, and the shapes of biological molecules such as **proteins**. Also a vital tool used in **pharmacogenetics**.

Carrier: A person who has one copy of a mutated **gene** causing an **inherited recessive disorder**. They are not affected but may pass the mutated gene onto their children.

Cell: The smallest unit of living organisms which, given the right conditions, can survive independently and reproduce itself. It has been estimated that the body of a human adult comprises 50 million million cells.

Chromosome: The self-replicating genetic structures of cells containing the **DNA** that bears the gene sequence. Each human **cell** normally has 46 chromosomes. 44 chromosomes are 22 matching pairs, where one chromosome of each pair is inherited from each parent. The other two chromosomes are the X and Y sex chromosomes. Normally, females have XX and males have XY.

Cloning: The process of asexually producing a group of cells (clones), all genetically identical, from a single ancestor. A clone is genetically identical to its parent and its siblings.

Cystic fibrosis: An **inherited recessive disorder** characterised by chronic infections of the respiratory tract, liver disease and digestive problems. Onset is usually in early childhood, and affected individuals require lifelong medical care. For those worst affected, cystic fibrosis is usually fatal by their early thirties.



Cytogenetics: The study of the structure of **chromosomes** and cell division. Cytogenetic tests detect chromosomal abnormalities or abnormalities in the number of chromosomes.

DNA (deoxyribonucleic acid) (also “Genetic code”): The double stranded helical chemical molecule that encodes genetic information. It is the code for life. The genetic code of nearly all living things is made of DNA.

Down’s syndrome: A genetic condition caused by the presence of an extra **chromosome** which is associated with certain physical characteristics and varying degrees of learning disability. Also known as trisomy 21.

False positive result: A positive genetic test result which, incorrectly, indicates the presence of a particular **mutation**.

False negative result: A negative genetic test result which, incorrectly, indicates the absence of a particular **mutation**.

Familial hypercholesterolaemia: An inherited disorder of fat metabolism giving rise to high blood cholesterol levels and a raised risk of heart attacks at an early age.

Gene: The fundamental physical and functional unit of heredity. A gene is a sequence of **DNA** that codes for one, or more, **proteins**. A virus such as HIV has under a dozen genes, bacteria can have about 5000 genes, yeasts can have about 7500, and humans have around 30000 genes.

Gene chip: (also known as DNA chips or microarrays): A small glass slide containing a matrix of different **DNA** sequences, representing many different **genes** in the **cell**, laid out in an array of spots. Used in research, for example, to measure which genes are switched on, or off, in a particular cell, and to detect **single nucleotide polymorphisms**.

Gene therapy: The deliberate introduction of genetic material into a patient’s **cells** in order to treat or prevent a disease.

Genetic conditions: Conditions which are direct consequences of defects in single **genes**; or in whole **chromosomes**, parts of which may be lost, duplicated or misplaced; or due to the interaction of multiple genes and external factors. Later in life such interactions appear to be the basis of many of the common serious disorders, such as heart disease, diabetes and cancer.

Genetic counsellor: In the UK, a health professional providing genetic counselling in a clinical setting. Genetic counselling is a communication process between the counsellor and the individual or family which deals with the medical and other implications associated with the occurrence, or the risk of occurrence, of a genetic disorder in a family.



Genetic test: A test to detect the presence or absence of, or change in, a particular **gene** or **chromosome**. This includes indirect tests which are not based on **DNA** but look for a product, such as a **protein**, indicative of a specific **gene** change in a person.

Genome: A person's genome is their total genetic information i.e. everything contained in their **DNA**.

Haemophilia: An **X-linked recessive disorder** in which the ability of the blood to clot is greatly reduced, causing severe bleeding from even a slight injury.

Huntington's disease: A hereditary disorder of the central nervous system characterised by uncontrollable irregular jerky movements and associated with progressive dementia. Although the genetic defect is present from birth, Huntington's disease is generally an **adult-onset disorder**, and is invariably fatal.

Inherited dominant disorder: A disorder where, for the person to be affected, the **mutated gene** only has to be inherited from one parent. Dominant disorders include **Huntington's disease** and **familial hypercholesterolaemia**.

Inherited recessive disorder: A disorder where, for a person to be affected, the **mutated gene** has to be inherited from both parents. Such parents are usually unaffected **carriers** because they only have a single copy of the affected gene. Recessive disorders include **cystic fibrosis** and **sickle cell disease**.

Muscular dystrophy: A number of related genetic diseases characterised by progressive deterioration of muscle shape and wasting of muscle. Duchenne muscular dystrophy is the commonest and most severe form. A similar but milder condition known as Becker muscular dystrophy is caused by different **mutations** in the same **gene**. Both are **X-linked recessive disorders**.

Mutation: The change in a **gene** or **chromosome** that can cause a disorder or the inherited susceptibility to a disorder.

Paternity test: A test which uses **DNA** analysis to determine whether a man is the biological father of a particular child.

Penetrance: The penetrance of a genetic **mutation** is the proportion of people with that mutation who develop that particular **genetic condition**. Penetrance is often expressed with reference to a particular age. For example, the penetrance of certain BRCA1 gene mutations (for breast/ovarian cancer) by age 70 has been estimated to be up to 85%.



Pharmacogenetics: The study of how people respond differently to drugs due to their genetic makeup, in terms of both how well the drug will work and what side effects the person might suffer.

PKU (Phenylketonuria): A metabolic disorder characterised by the abnormal accumulation of the protein component phenylalanine in the body fluids, resulting in severe mental deficiency. A low phenylalanine diet can prevent symptoms of the disease, and a screening test (the Guthrie test) is therefore carried out on all UK babies at age seven days. An **inherited recessive disorder**.

Predictive value: The **positive** predictive value of a test result is the likelihood that a person with a positive result will develop the condition. The **negative** predictive value of a test result is the likelihood that a person with a negative result will not ever develop the condition.

Protein: A large chemical molecule composed of one or more chains of smaller molecules in a specific order; the **gene** coding for the protein determines the order. Proteins are required for the structure, function, and regulation of the body's cells, tissues, and organs, and each protein has a unique function.

Sickle cell disease: A hereditary form of severe anaemia due to changes in the structure of the oxygen-carrying red blood pigment **protein** haemoglobin. The deformed haemoglobin causes red blood cells to become sickle-shaped in conditions of lowered oxygen, such as during infectious illnesses. The sickle shaped cells block blood vessels, cut off the blood supply to body organs, and cause painful sickle crises.

Single nucleotide polymorphism (SNPs): A variation in a single base (letter) of the DNA sequence which is present in different individuals and which may be associated with variations in susceptibility to diseases or the way that the body treats certain chemicals.

Thalassaemia: A hereditary disease resulting from defects in the synthesis of the oxygen-carrying red blood pigment **protein** haemoglobin. A type of anaemia.

X-Linked recessive disorders: Those disorders due to a mutation on the X chromosome. X-linked recessive disorders usually only affect males, but the disorders can be transmitted through healthy female **carriers**. Examples are **haemophilia**, **X-SCID**, and **muscular dystrophy**.

X-SCID: An inherited disorder affecting mainly boys in which the immune system fails to develop normally leaving the child susceptible to infections.





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

Online

www.tso.co.uk/bookshop

Mail, Telephone, Fax & E-mail

TSO

PO Box 29, Norwich, NR3 1GN

Telephone orders/General enquiries: 0870 600 5522

Order through the Parliamentary Hotline Lo-call 0845 7 023474

Fax orders: 0870 600 5533

E-mail: book.orders@tso.co.uk

Textphone 0870 240 3701

TSO Shops

123 Kingsway, London, WC2B 6PQ

020 7242 6393 Fax 020 7242 6394

68-69 Bull Street, Birmingham B4 6AD

0121 236 9696 Fax 0121 236 9699

9-21 Princess Street, Manchester M60 8AS

0161 834 7201 Fax 0161 833 0634

16 Arthur Street, Belfast BT1 4GD

028 9023 8451 Fax 028 9023 5401

18-19 High Street, Cardiff CF10 1PT

029 2039 5548 Fax 029 2038 4347

71 Lothian Road, Edinburgh EH3 9AZ

0870 606 5566 Fax 0870 606 5588

TSO Accredited Agents

(see Yellow Pages)

and through good booksellers

0 10 157912 8