

Addressing Genetics Delivering Health

A strategy for advancing the dissemination
and application of genetics knowledge
throughout our health professions



Public Health
Genetics

Hilary Burton

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Project Team

Dr Hilary Burton
Consultant in Public Health Medicine, Public Health Genetics Unit, Cambridge

Professor Janet Grant
Director, Open University Centre for Education in Medicine

Mr Mike Greenwood
Media Analyst, London

Ms Denise Jillions
Public Involvement Manager, Public Health Genetics Unit, Cambridge

Ms Helen Jones
Project Researcher, Open University Centre for Education in Medicine

Dr Mark Kroese
Specialist Registrar in Public Health Medicine, Public Health Genetics Unit, Cambridge

Ms Heather Owen
Senior Research Officer, Open University

Dr Alison Stewart
Chief Knowledge Officer, Public Health Genetics Unit, Cambridge

For additional copies please contact
Cambridge Genetics Knowledge Park
Strangeways
2 Worts Causeway
Cambridge CB1 8RN
Tel & Fax: +44 (0) 1223 740200

This report can also be downloaded from the following website: www.phgu.org.uk

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This project has been a huge team effort and alongside the sheer professionalism there has been much good humour and new friendships have been forged. I thank all my colleagues for this and look forward to continuing these relationships in future arenas.

Foreword

These are exciting times for genetics and the health service; rarely a day passes without announcement of some novel advance in genetics relevant to medicine. For some of these developments there is a way to go before their clinical benefits can be realised, but the impact of genetics on health is already being felt. As the former Minister of Health, Alan Milburn, said in 2001: “One thing is for certain: genetics will, indeed already is changing the world in which we live – holding out the potential for new drug therapies, new means of preventing ill health and new ways of treating illness”. The key significance of genetics in medicine was further underlined by the recent government White Paper *Our Inheritance, Our Future – Realising the potential of genetics in the NHS* (June 2003), which set out a commitment by the Department of Health to prepare the NHS for the genetics revolution, including an investment of £50 million over the next three years to help realise the benefits of genetics in healthcare in England.

Recognition of the importance of genetics to the health service last year led The Wellcome Trust and the Department of Health to commission the Public Health Genetics Unit to develop a strategy for genetics education in the NHS across the whole of the UK. With the increasing importance of genetics in all areas of health care, there is an urgent need to educate staff in the basic principles and applications of genetics. This document sets out the results of that process, developed with input from key stakeholders including specialist geneticists, those involved in clinical education and learning and the public. Through consultation with a wide range of health professionals for whom genetics is rapidly gaining prominence (including doctors, nurses, midwives and health visitors, dietitians, pharmacists and health service managers), Dr Burton and colleagues have produced evidence of the current state of genetics education and have developed with them an understanding of their further educational needs in this area.

Through the results of this excellent project, an educational strategy is now available for the dissemination and application of genetics knowledge throughout the UK, taking forward the vision of a workforce appropriately equipped to support patients by their understanding of genetic developments in healthcare. The task will not be easy, but the direction is now clear. It is my great hope that the communities will now work together to bring this to fruition.

Professor Neva Haites

Chair of British Society for Human Genetics

Summary of Recommendations

Establishment of a national Steering Group for Genetics Education

- drawn from many organisations with a UK wide role
- providing leadership and vision for the genetics education of health workers
- providing strategic overview of education programme
- collaborating with NCHPEG in the US
- ensuring continuing resources for the programme
- responsible for establishing and steering the Centre for Genetics Education

Establishment of a Centre for Genetics Education

- coordinating the education programme
- promoting genetics in NHS strategy and motivating its workforce to learn
- pressuring for inclusion of genetics in all relevant curricula
- seeking out new ways to develop genetics competence and share learning
- developing those professionals with a genetics special interest
- commissioning educational programmes using varied electronic resources

Establishment of a formal Programme for Genetics Education

- reviewing curricula at each stage of education and for each professional group: undergraduate and postgraduate medicine; nurse education; pharmacists; dietitians; public health professionals; health service managers
- facilitating the inclusion of core competency in genetics as a foundational component of all health training and education
- ensuring diverse, responsive, practical and relevant methods of teaching

Executive Summary

Introduction

The enormous international investment at the end of the last century in the Human Genome Project (HGP) underlined recognition throughout research and clinical establishments that genetics would transform our understanding of health and disease and, with it, the practice of medicine. As we move into the 21st century this becomes a reality as new technologies are developed that increase our capacity to predict, prevent, diagnose and treat disease.

Until recently, genetics was a relatively small specialty concerned with rare single gene and chromosomal disorders. The findings from the HGP, however, are challenging these boundaries and health professionals throughout the health service will increasingly be confronting new genetic technologies. Accordingly, an understanding of genetics needs to become a fundamental component of the scientific knowledge and practical competence of our healthcare workforce. This role, as key mediators ensuring that the outcomes of research can be realised through health benefits for society has been recognised as an important theme in The Wellcome Trust research programme and within NHS policy.

The identification and characterisation of more single gene disorders in many areas of clinical practice already provide a challenge to clinicians as they respond to concerns from patients about family history and steer them through decision making about testing and preventive services. Even this aspect of genetics is relatively unfamiliar at present to clinicians, most commonly emerging in the context of antenatal care and general practice. In the future, as genomic medicine becomes a reality, health professionals will be moving into uncharted territory. The identification of common gene variants associated with major chronic disease such as coronary heart disease, asthma, diabetes, psychiatric disease and cancers will lead to opportunities to test for increased disease susceptibility and offer prevention options or lifestyle advice. Furthermore, pharmacogenetic testing will be used to provide individualised treatments and genetic information from cells involved in disease processes will be used to provide greater levels of accuracy of diagnosis, and sometimes for screening.

There is much work still to be done to consolidate the research basis for many of these activities and there is debate about how long this will take. Nevertheless, there is little doubt that over the next decade or so, such possibilities will become a reality. Our capacity to reap the ensuing health benefits for the population will be dependent on having a workforce that can embrace these developments, bringing the full weight of their clinical experience into shaping and absorbing them into their practice. An increasingly knowledgeable public will demand no less.

Achieving this will require our health professionals to be equipped with the necessary educational background. Genetic science has progressed so rapidly during recent years that educational establishments and professionals in practice have not been able to keep up and a widening gap has developed. Change will not be easy.

The health care system is immensely complex and the use of new genetic applications will be very pervasive. So targeting any small group of professionals will not be an option. Rather, education in genetics needs to infiltrate the whole system, from undergraduate to continuing professional development and from practitioners in primary and community care to those in specialist practice. A substantial programme will be required.

This document sets out the basis of need and a strategic framework for just such an education programme.

Project

In 2002 the Department of Health and The Wellcome Trust commissioned the Public Health Genetics Unit (PHGU) to work with key stakeholder groups to develop a strategy for education in genetics for health professionals. This built on a background review commissioned by The Wellcome Trust and undertaken by the PHGU the previous year that had included needs assessment, educational methods, and a survey of current education¹. The report drew attention to the relative lack of genetics teaching within all areas of professional education. Any positive developments in the field seemed unsystematic and were usually products of local enthusiasm rather than any wider policy. Genetics teachers were scarce and most establishments could not cover the whole breadth from basic science to ethical, legal and social issues (ELSI). Evidence also suggested that educational approaches must be diverse, sustained in the long-term, and responsive to different learners' needs and varying access to resources.

In short, education in genetics for health professionals across the length and breadth of the UK had not kept up with scientific and clinical progress. But reversing this trend would be a difficult and time consuming process because of the unfamiliarity and breadth of genetics content, the complexity of both educational and health systems and the cumbersome mechanisms to influence them.

The project aimed to bring together a wide range of health professional groups, experts in genetics and experts in medical education and to explore with them the implications of genetics for practice in each professional group, their requirement for genetics education, the practical ways in which a broad genetic educational programme could be established for all health professions and the structures that would be needed to underpin this.

¹ Burton, H. Education in genetics for health professionals. Report to The Wellcome Trust. Public Health Genetics Unit. July 2002

Such a programme would need to:

- Be based on sound educational theory and practice
- Be supported by the genetics community whose expertise would be needed as genetics became more widespread in the NHS and who would be involved in much of the education
- Have support from the various stakeholders, including the professional associations and Royal Colleges; providers of education; professionals in practice; the public and people with genetic disease
- Tie in with existing educational structures and programmes without overburdening current curricula
- Be far-reaching, adaptable and sustained over many years as the clinical applications of genetics expand and are clarified and new roles develop

Method

The strategy development process was guided by a small steering group of individuals from the Department of Health and The Wellcome Trust, British Society for Human Genetics, Genetics Interest Group and the deans of UK medical and nursing schools. Educational expertise was provided by the Open University Centre for Education in Medicine.

Central to the process was a series of stakeholder workshops covering the following public and professional sectors: patients and carers, postgraduate medicine, nursing, midwifery and health visiting, primary care, pharmacy practice, dietetic practice and health service managers.

Workshops to which the public was invited were used to allow patients and carers to reflect on their experience of genetic disease and the requirements they had of health professionals. In professional workshops we brought together a breadth of interest including practitioners, teachers, professional organisations, researchers, and specialists in genetics. Structured discussions were used to obtain views on: the impact of genetics; awareness and readiness of the profession; opportunities and barriers to education. A nominal group process was used to obtain priorities for educational topics and resources required.

Further topics including undergraduate medical education in genetics, the use of UK genetics websites and the role of pharmaceutical companies in genetics education were pursued in depth through reviews, interviews, surveys and visits.

A final workshop was held in May 2003 in which all the material was presented and debate undertaken to provide the outline of the final strategic framework.

Results

The process provided a wealth of information which is published as separate reports on the PHGU website and summarised as evidence in the strategy document.

Work with the public and patients emphasised the point, accepted by professionals, that an educational programme must have the needs of patients and the public at its heart. As a fundamental principle the process of developing the programme must, itself, be publicly accountable. Education should prepare professionals for a future in which patients are better informed and expect to be partners with professionals in deciding on care. Finally, educational programmes should seek to utilise the power of first-hand accounts from patients.

The importance of genetics was agreed by all professional groups, but getting it onto the educational agenda will be an uphill struggle for all professions because of many competing priorities. This will be compounded by common professional perceptions that genetic disease is rare and that there is little current clinical utility. Whereas for undergraduates there should be a shift towards teaching of genomic medicine – the genetics of individual variation and common diseases - for today's practitioners education must be orientated towards the 'here and now' if it is to have any hope of achieving an impact. Priorities for primary care such as cancer genetics, antenatal and neonatal genetic screening programmes were agreed. For professionals in areas such as obstetrics, hospital specialties such as cardiology or neurology, pharmacy practice or those involved in chronic disease care, the content of genetics education would need to be carefully tailored to the particular clinical context.

The formal processes influencing professional education are powerful but can be hard and slow to change. Work must be done to develop and establish an understanding of competencies for all professional areas and levels of practice. This has already been achieved for undergraduate medicine with some effect in influencing the developing programmes. All professions need teaching in basic science, clinical aspects and ethical, legal and social aspects (ELSI) of genetics but it was considered that many higher education establishments will lack teaching capacity, and most will be unable to field the full range to include ELSI aspects. Our review also showed recognition that high quality educational material, such as that which includes first-hand content, or clinical scenarios, is time-consuming to develop. However, there is a broad willingness of institutions to share development and resulting resources.

For those in practice, incentives to genetics learning through such means as the inclusion of standards on genetics in National Service Frameworks and the promotion of education as an integral part of developments of genetics in clinical practice should be exploited. Learning should be made easy by the provision of a wide range of material accessible to people with different learning styles, and by the ready availability through the Internet of information, guidance and education in genetics to support everyday clinical practice.

Members of the final workshop recommended that ownership of the education programme should be consciously sought by involvement at three levels:

- Statutory bodies and professional associations
- Higher education institutions and other education providers and commissioners
- Local level including those in everyday practice

Responsibility for provision of this wide range of education would remain within normal mechanisms and would continue to rest with current educational providers. However, a special programme would be needed to provide the necessary energy to promote genetics in all fields, to undertake the necessary detailed work on educational resources and to coordinate work for maximum effect. Support might be achieved for such a programme from various sectors in the UK including major charities and the pharmaceutical industry and from the experience already gained in the United States.

Strategic Framework

The need for a strategic and wholesale approach to genetics education was strongly supported by all professional groups and, through the project, commitment was achieved to work together on a programme.

Based on the workshop and review findings and further discussion within PHGU and with experts in professional education, we make recommendations for the establishment and development of such a programme and its major elements.

We recommend the establishment of:

- (a) A national **Steering Group** for Genetics Education
- (b) A **Centre** for Genetics Education and
- (c) A formal **Programme** for Genetics Education

Establishment of a national Steering Group for Genetics Education

The establishment of a national Steering Group will be essential for championing the cause of genetics education amongst all professional staff and for bringing together all those with an interest in the development and/or provision of genetics education. With a membership drawn from many organisations with a UK wide role and, crucially, from the public and patients, it will:

- Provide leadership and vision for the genetics education of health workers
- Ensure continuing resources for the Programme
- Maintain a strategic overview of the education Programme
- Be responsible for establishing and steering the Centre for Genetics Education
- Collaborate with NCHPEG in the US

Establishment of a Centre for Genetics Education

A Centre for Genetics Education should be established that will have a remit to develop the UK Genetics Education Programme. The Centre should be developed as an enterprise of an established parent organisation such as a higher education institute, NHS trust, charitable or private organisation or a genetics knowledge park. Its eventual structure will need to take account of the needs of the UK as a whole.

It would have demonstrable expertise in:

- The education of health professionals
- The full breadth of genetics, including science, clinical aspects and ethical, legal and social aspects
- Networking with a wide range of partners
- Communication
- Database management, website development and management, and information technology

Establishment of a formal Programme for Genetics Education

We recommend that the components of the Programme for genetics education should include:

Leadership and coordination The Centre should achieve leadership and coordination for genetics education through its role as a focus for development that promotes and maintains an overview of initiatives and promulgates good practice.

Raising awareness and motivation to learn in practising professionals The Centre should pursue a programme to raise awareness of genetics amongst health professionals in three main ways: using formal opportunities; by building on needs arising through clinical practice; and by promoting its own work

Pursuing formal opportunities to promote genetics education in health service provision The Centre should pursue formal opportunities to promote genetics education within the NHS by identifying, promoting and coordinating involvement in national policy work and by promoting and contributing to the integration of genetics education as part of service developments

Promoting the development of leaders and facilitators with a special interest in genetics The Centre should promote the development of professionals with a special interest in genetics and support an educational role through formation of a network.

Developing core competencies in genetics The Centre should work with statutory and professional bodies and educational establishments to ensure that:

- Genetics education is embedded in general education at undergraduate, professional, specialist and continuing development levels as appropriate for that profession
- Consideration is given to training and accrediting physicians, nurse and others with a special interest in genetics

Development of educational programmes A rolling programme of educational resource development should be implemented covering each professional group and all levels based on an understanding of needs and priorities. Each area would need input from a range of experts and, importantly, the public and patients with genetic disease.

Facilitation and sharing of resources The Centre should promote and facilitate the sharing and dissemination of educational resources through the development of a database and information service

The development of electronic resources to provide clinical support and information A major programme should be developed to provide access to authoritative information, clinical support and educational resources via the Internet. This should include further development and functional linking of the NeLH genetics site, BSHG and regional genetics centre websites and provision of links to specific educational material.

Implementing the Genetics Education Programme

The resources to set up such a Programme would be substantial. Success will only be achieved by the establishment of a significant presence and the completion of highly regarded work. Resources to set up and run the Centre alone will be in the order of £750K annually and further funds will be needed to develop educational materials and to pump-prime other developments such as intensive 'training the trainer' programmes.

All in all, we estimate that an annual budget of around £2 million will be required. Ideally the commitment for this work should come from a range of bodies including the Department of Health and other major research organisations and charities, as well as the private sector. In supporting this work they would take their share of responsibility for the preparation of the NHS workforce in genetics, an outcome that is much in their interests. They will also ensure the best possible chance that the ensuing educational developments will be rapid, high quality and achieve maximum coverage.

Conclusion

We believe that the time has now come to implement our recommendations and, for this, the announcements in the Genetics White Paper provide a very welcome catalyst. We should move forward in a partnership that acknowledges the interests of the many organisations in this process, from the researchers keen to see the translation of their work into health benefits to those involved in the delivery of health services, and from public health specialists to the private sector.

In 2001 the Secretary of State announced a new ambition for Britain: to put us at the leading edge of advances in genetic technologies and to develop in the UK a modern genetics health service unrivalled in the world. A competent workforce will be fundamental to achieving that vision.

Section One

Introduction, Project Outline and Strategic Framework

I Introduction

I.1 Context

The enormous international investment at the end of the last century in the Human Genome Project (HGP) underlined recognition throughout research and clinical establishments that genetics would transform our understanding of health and disease and, with it, the practice of medicine. As we move into the 21st century this becomes a reality as new technologies are developed that increase our capacity to predict, prevent, diagnose and treat disease. Alongside this process of scientific discovery the major research programmes showed great foresight in recognising the huge impact that genetics would have on society. A programme on ethical, legal and social implications (ELSI) was set up as an integral part of the Human Genome Project to foster basic and applied research and support educational outreach.

One of the most important ways in which this impact will be mediated is through the integration of genetic technologies and information into healthcare and public health activities. For this, an educated and competent health workforce will be required - a prerequisite noted by The Wellcome Trust within its overall programme concerned with public interest in science and biomedical ethics, and by the Department of Health, most recently in its White Paper², published in June 2003, *Our Inheritance, Our Future*. In this document the government signalled its intention to prepare the NHS workforce through a major new programme to 'spread knowledge' throughout all sectors.

In recognition of the importance of genetics education for health professionals, in July 2002 the PHGU was commissioned jointly by these two organisations to work with professional groups on the development of a framework for a national educational programme in genetics for health professionals. This document represents the culmination of that process.

I.2 Background

The framework builds on a review commissioned by The Wellcome Trust undertaken by the PHGU the previous year³. This had shown that genetics education was not a high priority within the broad range of NHS policy. Nor was it widely accepted as important by health professionals, although in each professional group there were individuals who had realised the likely future impact and were calling attention to the necessity of preparing the workforce.

² Our Inheritance, Our Future - Realising the potential of genetics in the NHS. The Stationary Office. June 2003. The White Paper is also available at www.doh.gov.uk/genetics/whitepaper.htm.

³ Burton H. Education in genetics for health professionals. Report to the Wellcome Trust. Public Health Genetics Unit. July 2002

Educational programmes from undergraduate to continuing professional development (CPD) were variable in their genetics content with no professional groups systematically covering the full range from basic science to ethical, legal and social issues in their curricula. Whilst there were some developments across the country such as new modules developed by some universities and nursing schools, these were piecemeal and products of local enthusiasms and interest, rather than systematic. Teachers in genetics were scarce, with specialists in regional genetics departments being overstretched as they attempted to bring genetics to a wide range of their local professionals. The newly established genetics knowledge parks all had a commitment to education, with varying focus on professionals and the public, and were beginning work in this area.

The background report also looked at the effectiveness of educational approaches with health professionals, concluding that there was no good evidence that any particular methods were superior. Rather, the report found that a multiplicity of methods was required, sustained over a period of time, to suit people with different learning styles and variable access to educational resources. The Internet was a valuable source of education, with a wide range of educational material available worldwide, but it was not clear how well this was set up in the UK to provide for professionals' needs for information and education in support of clinical practice. Some regional medical genetics departments had attempted to do this through websites but, again, this was neither systematic nor necessarily based on an understanding of the needs of the user.

The report concluded that a strategy for education should be developed to bring together those with an interest, whether as commissioners or providers of education, in order to set out a clear vision and prioritise future work. To achieve this the main strategic groups would need to undertake further work in those areas that would be necessary to inform decisions.

This framework is the result of that process. It sets out the main findings and makes recommendations for the establishment of a national Steering Group, a Centre for Genetics Education and the setting up of a formal Programme for developing genetics education in the health workforce.

1.3 The strategy-building process

From the background report we knew that we would have to consider:

- A wide range of professional groups
- Individuals at different levels of education
- The requirements of different specialities

The eventual programme would need:

- To be based on sound educational theory and practice
- To be supported by the genetics community whose expertise would be needed as genetics became more widespread in the NHS and who would be involved in much of the education

- To have support from the various stakeholders, including the professional associations and royal colleges; providers of education; professionals in practice; the public and people with genetic disease
- To tie in with existing educational structures and programmes without overburdening current curricula
- To be far-reaching, adaptable and sustained over many years as the clinical applications of genetics expand and are clarified and new roles develop

In an area where there were few specialist teachers and many needing to learn, the challenge would be to use all our expertise effectively and efficiently to develop programmes and materials that could be widely accessible. We would strive to use all available methods to 'drip-feed' genetics and genetics concepts into professional practice within the NHS, and to ensure that the developing programme would have the confidence of patients and the public.

1.4 Aims of the project

The project aimed to bring together a wide range of health professional groups, experts in genetics and experts in medical education and to explore with them:

- The particular issues raised by genetics for each professional group and the degree to which this was recognised by individuals and organisations
- Their need for genetics education based on needs of patients with genetic disease and the wider public
- The practical ways in which a broad genetics educational programme could be established for all health professions and the structures that would be needed to underpin this

Whilst recognising that education in genetics would remain the responsibility of a wide range of providers as well as the individuals themselves, two key questions remained: firstly, how to raise awareness and motivate learners and, secondly, how to gradually develop competence through stimulating, moulding and shaping the existing formal and informal mechanisms.

2 Methods

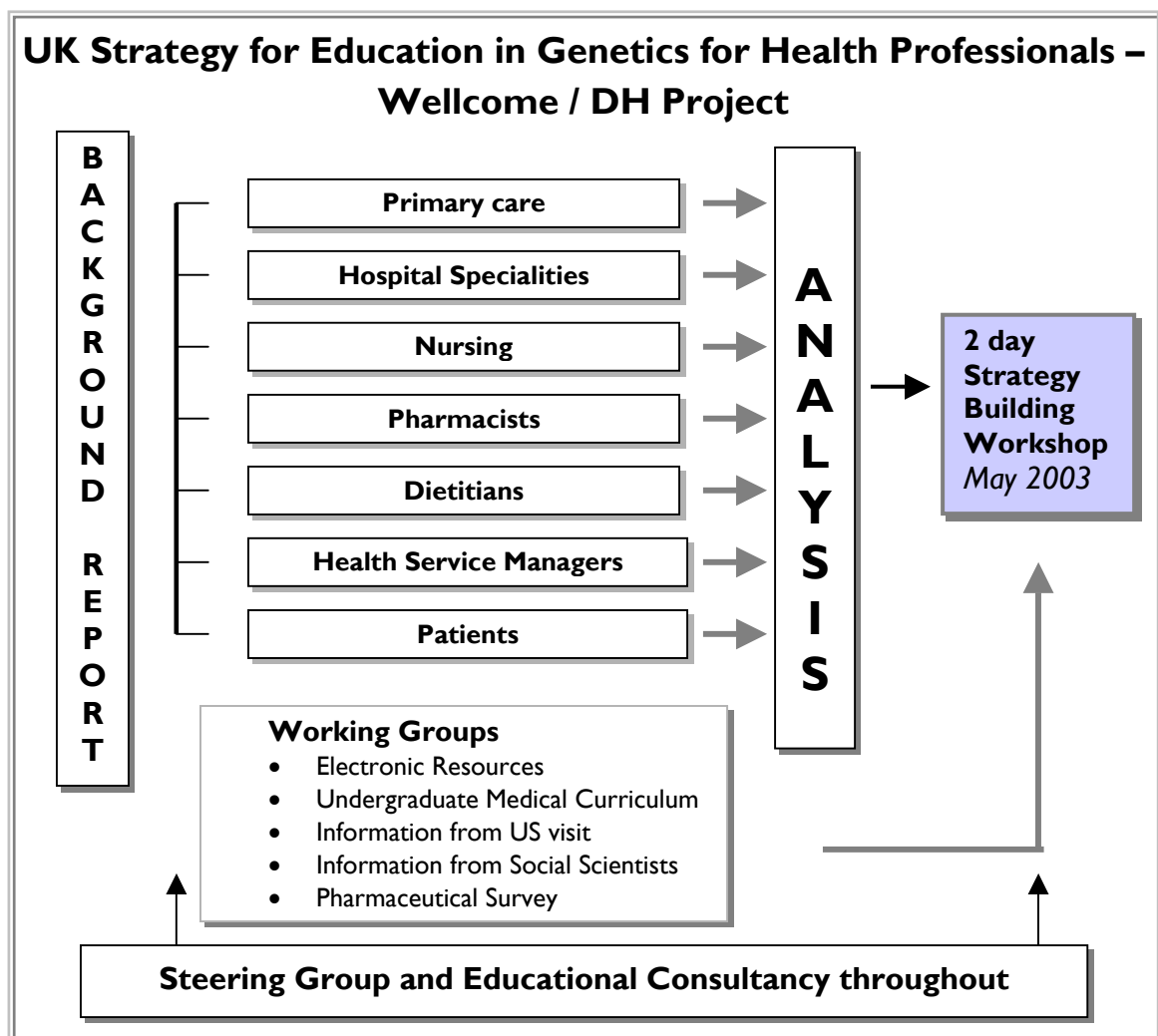
2.1 Introduction

The project was wide in its scope, covering the educational needs of five main professional groups, the settings of primary and secondary care, the whole range of education from undergraduate to continuing professional development and the whole of the UK.

Professional groups included:

- Doctors
- Nurses, midwives and health visitors
- Pharmacists
- Dietitians
- Health service managers

The process involved many elements including a series of stakeholder workshops and a number of commissioned reviews. These took place between November 2002 and May 2003. A summary diagram is given below.



2.2 Steering group

The strategy development process was guided by a small steering group with membership from the Department of Health, The Wellcome Trust, the Public Health Genetics Unit, the Genetic Interest Group, the Conference of Postgraduate Medical Deans of the United Kingdom (COPMeD) and the Council of Deans and Heads of UK University Faculties of Nursing, Midwifery and Health Visiting. This group met twice to discuss the overall direction and content of the work, and the design of stakeholder workshops and the final workshop.

2.3 Educational expertise

A good understanding of the current climate for professional education, of available structures through which change would be implemented, and of the ways in which learning can be promoted at various educational levels was central to the work. The Open University Centre for Education in Medicine, led by Professor Janet Grant, was commissioned to provide educational expertise. Members of the Centre advised on the participants and programme for each workshop, participated in and observed each workshop and undertook a survey of undergraduate medical education in genetics throughout the UK.

2.4 Consultation visit with NCHPEG organisation in United States and attendance at NCHPEG conference January 2003

A team of four members involved with the project attended the conference of the National Coalition of Health Professionals Education in Genetics in Washington in January 2003. Members of the group were:

- Hilary Burton (Public Health Genetics Unit)
- Jon Emery (Department of Public Health and Primary Care, University of Cambridge)
- Alison Hill (Department of Health)
- Roni Liyanage (The Wellcome Trust)

Prior to the conference the group was able to have a long discussion with the NCHPEG Director, Joseph McInerney. This focussed on factors that were critical to the setting up and running of the organisation. Hilary Burton also attended the NCHPEG Board Meeting, chaired by Frances Collins. Members of the group attended all sessions of the conference. Opportunities to meet other delegates, particularly those concerned with education in primary care and nursing, had also been arranged for members of the group. The group presented papers at an evening workshop focussed on developments in genetics education outside the United States.

2.5 Stakeholder workshops

(a) Workshops for patients and carers

The Genetic Interest Group (GIG) led these workshops for the Public Health Genetics Unit (PHGU) working closely with Denise Jillions, the Public Involvement Officer at the Cambridge Genetics Knowledge Park (CGKP). It held two workshops

during March and April 2003 in London for patients with genetic disease and their families and carers. These workshops were both attended by members of the PHGU project team.

Participants were invited to reflect on their experiences of care from professionals following the patient's journey from initial awareness of disease, through diagnosis, to subsequent treatment and continuing care.

Findings from the two workshops were written up as a report that was then made available for comment both on the PHGU website and through the Human Genetics Commission's Consultative Panel, a panel made up of 106 people with direct experience of living with genetic disorders. The report was made available to all stakeholder workshops and the final document can be accessed via the PHGU website at www.phgu.org.uk.

(b) Professional workshops

Six professional workshops covered the following main areas:

- Postgraduate medicine
- Nursing, midwifery and health visiting
- Primary care
- Pharmacy practice
- Dietetic practice
- Health service management

To ensure ownership each workshop was planned by a small working group involving key members from that professional area. Through this we outlined the key issues for that profession and identified participants to advise from the point of view of education providers, those in practice, those who had a special interest in the relevance of genetics to the profession and, where possible, those involved in learning. As far as possible, membership was chosen from Scotland, Northern Ireland and Wales as well as from England.

Each workshop was attended by 8-12 participants drawn from the professional group as well as a clinical geneticist, and a representative of the Genetic Interest Group. The workshops were facilitated by team members from PHGU and the Open University. Colleagues from Department of Health and The Wellcome Trust were invited to observe any of the workshops and were able to attend at least part of most of them.

The workshops all went through a similar structured format designed to gather information on implications of genetics for the professional group, key issues, and a nominal group process through which clinical and teaching priorities and resources requirements were agreed (see Appendix 2 for further details). Full reports of each workshop are available on the PHGU website at www.phgu.org.uk.

2.6 Changing public expectations of the relationships with health professionals

A seminar to develop a broad context for strategic planning of genetics education was held with social scientists from the Centre for Family Research, University of Cambridge. The process was led by Denise Jillions, the Public Involvement Officer at the CGKP. A report of these findings was given to the final workshop and is available on the PHGU website www.phgu.org.uk

2.7 Review of websites provided by specialist genetics services in UK for clinical support, information and education

A review was commissioned from Mike Greenwood, an independent media analyst, to assess the role of online resources provided for professionals by regional genetics services and other specialist genetics organisations in the UK in giving access to information, clinical support and education. The review sought to identify the needs of health professionals, the kinds of online content which best met those needs, and to consider gaps in the current provision. The work included:

- An overall summary and critique of these UK sites
- An in-depth user-study of the West Midlands site conducted with a range of professionals involved with the provision or use of the site
- A series of interviews with people responsible for planning and creating specialist sites including the British Society for Human Genetics, the National electronic Library for Health (NeLH), the West of Scotland Regional Genetics Service (Institute of Medical Genetics, Yorkhill Academic Campus, Glasgow) and the East Anglian Medical Genetics Service (Addenbrooke's NHS Trust, Cambridge)

The findings from this review were presented to the postgraduate medicine, primary care and managers workshops. At the final workshop the report formed the basis for further discussion on the way forward for electronic resources to support clinical practice in the UK. The report is available on the PHGU website at www.phgu.org.uk.

2.8 Survey of education in genetics at UK medical schools

The Open University Centre for Education in Medicine, led by Professor Janet Grant, undertook a questionnaire survey of genetics teaching in all established medical schools in the UK. The survey was based on learning objectives defined by Professor Neva Haites of Aberdeen University, working with a professional group drawn from the British Society for Human Genetics⁴.

The survey sought to find out from genetics leads what happens now in medical schools' genetics teaching and what assistance, support and resources were needed for genetics education. This detailed study provided an example of how learning

⁴ Haites, N et al. Teaching Medical Genetics to Undergraduate Medical Students. 2002 Document agreed by British Society for Human Genetics and Joint Committee on Medical Genetics available at [\[www.bshg.org.uk/Official%20Docs/UNDERG~1.doc\]](http://www.bshg.org.uk/Official%20Docs/UNDERG~1.doc)

objectives in genetics could be embedded into an undergraduate curriculum, the educational resources that had been developed, further resources that were required, and the potential for future sharing. A report of that work was presented at the May workshop and the full report is available on the PHGU website at www.phgu.org.uk.

2.9 Survey of educational work in genetics undertaken by the pharmaceutical industry

A survey was commissioned from Cohn and Wolfe Policy Healthcare, an independent company with expertise in relations with the pharmaceutical industry, to investigate the current and possible future roles of the pharmaceutical industry in providing or supporting genetics education for health professionals. The survey was undertaken by Anne Ruglys and involved in-depth interviews with seven major pharmaceutical companies.

The interviews were aimed at finding out their infrastructure for education, their general educational activities, including any undertaken with public and patients, health professionals and schools and any specifically orientated around genetics. The survey sought information on their attitude to genetics education of health professionals, whether they regarded it as important, what skills they thought were required, whether they had any plans to develop this within the UK, and whether they would be prepared to be involved in developing a coordinated approach within the UK.

Results from this survey were presented at the final workshop and the report is also available on the PHGU website at www.phgu.org.uk.

2.10 Final strategy building workshop

A two-day strategy building workshop was held in May 2003 at Hinxton Hall, on The Wellcome Trust Genome Campus, Cambridge. This workshop was an opportunity to bring together representatives from each stakeholder workshop, the genetics community, the genetics knowledge parks, Genetic Interest Group, workforce development confederations and those interested in genetics services from the devolved administrations. The workshop included a chance to review all the material from the stakeholder workshops, and to hear presentations on the findings of the supplementary studies.

In preparation for the workshop, a small working group including team members from Open University, Department of Health and The Wellcome Trust met together to decide on the main outstanding issues for the programme. These were based on the outputs of the stakeholder workshops and resulted in a number of topics which formed the basis for small group discussions. See Box 1 overleaf.

Box 1 Discussion topics for final Hinxton workshop

Awareness and motivation to learn about genetics in healthcare. *How can we generate demand for educational development?*

Ownership. *How do we ensure ownership of educational programmes at national and local level?*

Internet resources. *How can we develop good Internet resources to provide information, clinical support and education in genetics for health professionals?*

Service developments. *What service developments are occurring or need to occur that have implications for genetics education? How can we build on these to understand in more detail what professionals need to know and how this can be provided?*

Central support and coordination. *How can central support and local coordination best be provided. What would be the functions of such support and what would be its necessary activities?*

The findings of these sessions are presented in appropriate sections of this report and the discussion at the workshop provided the basis of the eventual strategic framework.

3 Results

3.1 Introduction

A wealth of material was developed in the form of workshop reports and reports of further reviews. These are available in full on the PHGU website at www.phgu.org.uk and are collated and summarised in Chapters 5-19. They provide the detailed evidence for the resulting **Strategic Framework** which is set out in the next chapter.

In the Strategic Framework we aim to build on current understanding of what is needed by patients and the public both now, and as genetic science progresses over the next five to ten years. Our starting point for education is the current awareness of genetics and the recognition amongst health professionals of a need to learn. An educational programme must fulfil these needs whilst at the same time anticipating future needs and thereby preparing professionals to embrace further developments arising from genetic science. It must also understand the competencies that professionals need to acquire and the ways in which they learn at all levels, ensuring that a wide range of resources is developed and made available to meet these learning needs in an effective and efficient manner.

The evidence in support of the framework is set out under the following chapter headings and summarised here as some of the main emerging policy points.

3.2 An evolving public

With better access to information and increasing demand for accountability, the public will expect to be involved in decision making about major health service developments. In genetics this will be against a backdrop of growing public interest and, sometimes, disquiet about the scientific developments and their applications.

Individual practitioners will be operating in an environment that increasingly acknowledges that the special understanding and insight gained by experience of illness is a valid form of expertise (the 'expert patient').

As a direct implication of this, public and patient consultation, and indeed involvement at every stage, must play a primary role in the development of the education strategy.

3.3 The patient perspective

The needs of patients must predetermine the development and design of the service they receive. This will require an interrelated patient-professional framework to structure and inform the content and context of the education of the health professional. Patients and carers should be involved in all aspects of educational provision from development, through to delivery and assessment. Sourcing the information with which to develop such educational programmes should focus on first-hand accounts, not only as resource material but also as high impact educational tools. Whilst we seek to promote an effective utilisation of real experiences in this way, we also stipulate the need for accompanying guidelines that will implement the

necessary safeguards, ensuring we learn and teach from the experience of patients without any risk of their exploitation.

3.4 Awareness of genetics

There is a lack of awareness amongst health professionals about genetics that will prove an obstacle for each professional group. There is a consensus that any approach must not only be highly relevant and accessible but also based on the current clinical utility of genetics knowledge within each profession. However, competence in genetics must be understood as a fundamental component of a comprehensive professional knowledge, not the result of chance personal interest, and accommodated accordingly within every aspect of the healthcare environment.

We recommend a wide-ranging and formal promotion of genetics awareness from strategic policy review and inclusion in government health standard requirements to the stimulation of media interest and by implication, through genetics-informed patients' raised expectations of their health professional. Thus, by policy, by exposure and by demand, we hope to motivate professionals to realise the potential for genetics in their field.

3.5 Priority areas for genetics education

As well as developing basic education courses to underpin future understanding, all professional groups have proposed priority areas where immediate education in genetics is required. These include:

- Cancer genetics
- Genetics as component of disease in other hospital specialties
- Antenatal and neonatal genetic screening programmes
- Obstetrics (pre-implantation, antenatal and neonatal genetic testing)
- Pharmacogenetics
- Chronic disease care for people with genetic conditions
- The development and management of specialist genetics services
- The implications of genetics for public health practice

3.6 Current opportunities

The relevance of genetics should be systematically considered in each National Service Framework by the inclusion of specialists in genetics and public health genetics in the working groups. Importantly, standards for care should require that health professionals are competent to deal with genetic aspects of disease.

A genetics education programme will require a thorough understanding of, and close cooperation with, emerging NHS educational structures including those involved in NHS University, Postgraduate Medical Education and Training Board, the developments of Practitioners with Special Interests, and systems of personal development, appraisal and review.

3.7 What do professionals need to learn?

Education in genetics must be based on learning needs arising from the current realities of professional work, rather than on future or conceptual possibilities. Although the professional groups place emphasis on different education content, all clinicians include in their learning needs at least some molecular biology, clinical skills including family history taking and risk communication, details of how to get further information on genetics and ELSI aspects.

3.8 Genetics and genomics: preparing for genetic information in health and disease

The emphasis in basic genetics education for all professionals should eventually shift from medical genetics and single-gene disorders to one that stresses the genetics and molecular biology underlying human variation and disease. Priority must also be given to the development of skills in risk assessment and communication with a view to incorporating genetics information in the provision of comprehensive health services when supported by the appropriate evidence.

Current computer assisted programmes for assessing risk and giving advice should also be promoted so that professionals can gain experience of new ways of working in advance of the emergence of new technologies. These must be accompanied by appropriate education and support.

In addition, all professionals should receive education in evidence-based medicine. In each profession some individuals should become more expert in the issues arising from genomics and in the skills required for critical appraisal in genomics in their special area.

Furthermore, as new applications incorporating genomics become available, these will need to be supported by the appropriate education for professionals in practice based on an assessment of their need. The professional will need clear information to support use of the applications and to know how to access and use the information.

3.9 Resources for formal learning

The development and implementation of genetics education for health professionals will require substantial resources and the advancement of teachers as well as systematic support for learners and local facilitation. Both learners and teachers should have access to high quality and diverse learning materials including first-hand accounts, scenario workshops and video and library resources. We are keen to emphasise this diversity, indeed, the methods of teaching must be responsive to the needs of every learner, and especially, must be rooted in the tangible benefits at the sharp end of health service delivery. This strategy should be developed at a national level, and incorporated in formal and clinically-based education schemes whilst being completely accessible to everyone.

3.10 Survey of medical undergraduate education

There is much support from professional groups for developing and sharing complex educational materials on a national basis, and for some consistent, centrally resourced method of doing so. The agreement, now achieved, on essential core knowledge and skills for medical genetics in undergraduate medical education should form the basis for any subsequent influence upon curricula as well as being the catalyst for future developments. Crucially, any additional responsibilities given to teachers must be accompanied by an equal increase in resource provision.

3.11 Learning in a professional environment

The inclusion of genetics in CPD is determined by the perception of the subject as important and useful, and as such, a worthy component of high quality services. An educational programme must look to create demand for genetics-based self-development in the professionals themselves. Importantly, the CPD programme for genetics should work through a range of learning methods from academic activity and professional meetings to healthcare practice and technological resources. It must also ensure provision for easy access to authoritative information, both written and from colleagues, and those with more experience.

3.12 The development of UK Internet resources to support clinical practice

The Internet will be a valuable resource for clinical support and education and the regional genetics departments, NeLH and other specialist genetics websites should be developed to provide clinical support, information on genetics diseases, and information about services for all health professionals as well as access to genetics education on the worldwide web.

3.13 The use of service developments to support clinical practice

There are many current and future expected applications of genetic technologies and, as these are developed and implemented, they will require parallel consideration of the accompanying educational requirements of the workforce. Opportunities should be systematically sought to tie genetics education to both current and future service developments where genetics is a significant component of care pathways.

3.14 Developing ownership for the genetics education programme

Ownership of the education programme should be sought throughout the service, including:

- Statutory bodies and professional associations
- Higher education institutions and other education providers and commissioners
- At local level including those in everyday practice

3.15 The role of the pharmaceutical sector

The importance of and need for genetics education for health professionals was endorsed by our survey of the pharmaceutical sector. All were willing to participate

further in discussions to develop a co-ordinated approach and we recommend that partnership from the pharmaceutical sector is sought in the development of an educational programme.

3.16 Experience from North America

The UK should seek alliance with NCHPEG in developing its educational programme in order to build on the work already achieved in the US and in the longer term to work collaboratively on the development of further educational resources.

In the following chapter we bring together these findings and, using these and the further work of the final strategy-building process, make recommendations for a **UK Strategic Framework for Genetics Education**.

4 A UK Strategic Framework for Genetics Education for Health Professionals

4.1 Introduction

Representatives of a wide range of health professionals have, for the first time, come together through the strategy building process to discuss their common interests in genetics education. They have all agreed the importance of developing in the NHS workforce a competence in genetics in order that they might respond to the transition of genetics from specialist to mainstream services and to the development of new technologies and interventions based on genetics and molecular science.

Since the completion of the workshops, the government has published its White Paper on Genetics, setting out its commitment to ‘spreading knowledge’ across the NHS and outlining a major programme of investment in England including the establishment of an NHS Genetics Education and Development Centre. Because many of the issues are the same, and because of the UK-wide remit of many of the professional and educational organisations, we would recommend that this programme should ideally cover the UK as a whole. Our workshop and review process, which has the support and commitment of the various health professionals, strongly supports this vision. We also suggest that the NHS Centre can only form one of many components of an education strategy, albeit an important central component. It is for this reason that we have separated our recommendations into three complementary strands.

The way in which genetics underpins so many aspects of health and healthcare means that the development of educational programmes for the NHS will be an extensive challenge, unprecedented in its scale and complexity. Amidst wide-scale lack of awareness of genetic science amongst the NHS workforce, and a growing conviction from scientists and professional experts that this will have a large impact on health and health care, we believe that there is currently a window of opportunity to prepare health professionals.

There is much debate and disagreement about timescales, and when genetics will have a major impact on healthcare. More likely than not, in spite of piecemeal advances, significant and radical change will be some years in coming, but the notion that changes will eventually arrive is not in doubt. The need for a strategic and wholesale approach to genetics education is therefore strongly supported by all professional groups. Its aim will not only be to anticipate the future impact of genetics across the whole of medical practice, but also to ensure that the here and now of the practice of medical genetics, and how that might affect diagnosis and treatment across a range of other specialties, are understood by the workforce as a whole. The only way that this can be achieved is through a proactive programme of education, centrally coordinated, but delivered and owned locally by a variety of professional groups.

We set out here our recommendations for the establishment and development of such a programme and its major elements. These are based on the findings of the workshops, and on further detailed consideration by the PHGU involving also the educational expertise of Professor Janet Grant of the Open University Centre for Education in Medicine.

We recommend the establishment of:

- (a) a national **Steering Group** for Genetics Education
- (b) a **Centre** for Genetics Education and
- (c) a formal **Programme** for Genetics Education

4.2 A Steering Group for Genetics Education

The Steering Group will comprise those who have an interest in the development and/or in the provision of genetics education for health professionals. These might include representatives of:

- Patients and the public
- The Human Genetics Commission
- The Genetic Interest Group and constituent voluntary organisations
- The Department of Health and the devolved administrations in UK
- Strategic health authorities and primary care trusts
- Research bodies such as MRC and The Wellcome Trust
- Industry
- Professional organisations such as The British Society for Human Genetics
- Statutory bodies such as the General Medical Council, the Nursing and Midwifery Council and the Postgraduate Medical Education and Training Board
- Royal colleges and related committees such as the Joint Committee on Medical Genetics of the Royal College of Physicians
- Higher education institutions
- Workforce development confederations
- Genetics knowledge parks

We take the view (acknowledging that it was not fully endorsed by the workshops) that the establishment of a Steering Group will be essential for championing the cause of genetics education among all professional staff and for maintaining an overview of the educational programme.

The chairmanship of the Steering Group will be a very influential role and should be invested in an individual who can bridge the research and health professional communities and who has the ability to inspire both. The Steering Group, drawn from many organisations, will in effect take responsibility for the strategic leadership of genetics education across the UK. It will have a UK wide role that would include:

- Leadership and the establishment of a vision for genetics education of health professionals across the UK
- A strategic overview of genetics education
- Collaboration and joint work with the National Coalition for Health Professionals Education in Genetics in the USA
- Work with other partner organisations with interests in genetics education
- Ensuring substantial and continuing resources for the Programme
- The establishment and subsequent steering of a Centre for Genetics Education

4.3 A Centre for Genetics Education

The development of a Centre for Genetics Education was again not an explicit recommendation of the workshops, but the idea of a driving force was a recurring theme. Set alongside the expressed need for a number of detailed functions, such as those of awareness raising, coordination and programme development, we concluded that some sort of physical reality would be necessary. However, the eventual nature of such an organisation, particularly the way in which the structure might relate to the needs of the UK as a whole, and to the proposed NHS Genetics Education and Development Centre would need further debate.

We envisage that the roles of a Centre will be to:

- Provide leadership in the development and coordination of the genetics education programme
- Promote the importance of genetics in NHS strategy
- Raise awareness and motivate health professionals throughout the NHS
- Exert pressure on professional organisations, societies and educational institutions to include genetics in their curricula
- Provide a focus within and between professions to ensure each can learn from the others
- Seek out and promote opportunities to develop genetics competence
- Promote the development of leaders and facilitators with a special interest in genetics across a range of professions and specialties
- Coordinate and commission the development of educational programmes including the use of web-based materials
- Facilitate the sharing of educational materials
- Be involved in the development of electronic resources for clinical support and information

We thought it useful to bring together views from the project about the functions that were needed, to think through further detail on the likely activities of the Centre and from here, to determine what expertise, staff and other resources would be required. These are set out in the Strategic Framework as examples that provide some idea of the possible roles and nature of the envisaged Centre and an approximate estimate of the level of funding that would be needed. The areas of work will provide the main strands of the Genetics Education Programme, which is outlined further in section 4.4.

What will the Centre for Genetics Education be like?

The Centre for Genetics Education will be a vibrant nerve centre for the development of education in genetics and will provide the energy and driving force for the development of a Genetics Education Programme across the whole of the UK.

The Centre is likely to be developed as an enterprise by an established parent organisation such as a higher education institution (HEI) or NHS Trust; a charitable or private organisation could also be considered. Given the wide remit of the genetics knowledge parks, and their focus on education, these might also be suitable organisations in which to host such a Centre.

The Centre would have demonstrable expertise in:

- The education of health professionals
- The full breadth of genetics, including science, clinical aspects and ELSI
- Networking with a wide range of partners and communication
- Database management, website development and management, and information technology

Some suggested functions of the Centre are given in Box 2.

Box 2 Centre for Genetics Education: some possible functions

- *Liaison with professional and academic bodies, agencies and providers of learning and the Workforce Development Confederations*
- *Being a pressure group that will help to promote the importance of genetics education in national policy areas such as the National Service Frameworks*
- *Ensuring that learning needs are understood for different professional groups and at different levels, and that skills and competency frameworks and appropriate curricula are developed*
- *Developing a network of providers of genetics education to facilitate sharing of resources and promotion of good practice. This will include the development of a website and communication functions such as a newsletter*
- *Collecting, reviewing and providing a database of electronically accessible learning support materials*
- *Commissioning new learning resources according to an agreed programme of priorities*
- *Providing an information service for education providers*
- *Running conferences, seminars and workshops, including occasional forums with special groups such as the business community*
- *Maintaining an overview of research in genetics education and carrying out evaluation and monitoring of educational resources*
- *Being involved in commissioning information and education resources to support clinical practice, such as regional genetics department websites, the NeLH and other information support systems*

To have the necessary impact the Centre will need its own offices and facilities, and a core of staff with expertise in genetics, education, IT, website development and

maintenance, project management and liaison (for example with genetics service development work), information services and communication. It will need to be supported by administrative and clerical staff.

It will also have a substantial capacity to commission expert involvement where required in the development of priority programmes.

It will have the capacity to develop educational material based on the content written by expert groups, either in-house, or in conjunction with another organisation.

It will have access to the necessary facilities and administrative support to put on conferences, workshops and other educational events.

4.4 The Genetics Education Programme

The Genetics Education Programme will be formally established by the Steering Group and should encompass priority areas described below as agreed during the workshop and review programme. Over forthcoming years the Steering Group will be responsible for review and refinement of the strategy. Most of the implementation will fall under the responsibility of the Centre.

I Leadership and coordination

Many eminent organisations have begun to shape the educational agenda for genetics. The Joint Committee on Medical Genetics, the Royal College of Nursing, The British Society for Human Genetics, the Public Health Genetics Unit and the Virtual Genetics Group of the Royal College of General Practitioners have together achieved a great deal of progress across many important issues in genetics. They have raised awareness among professional organisations, developed outline curricula for undergraduate and postgraduate physicians, and established educational programmes for policy makers and public health professionals. However, none of these has education in genetics as its main brief or core purpose. None has had the explicit remit to work with the full range of health professionals, such as pharmacists, dietitians and health service managers; nor have they had the authority, commitment or resources to drive the entire programme forward.

There have been a number of developments both formally and through individual innovative schemes, but provision has by no means been universal. Developments have been piecemeal and have often tended to collapse when funding is terminated. These initiatives require coordination so that educational programmes can be developed efficiently, evaluated and, if successful disseminated as examples of good practice.

We recommend that the Centre should achieve leadership and coordination for genetics education by ensuring a focus for development in one organisation that promotes and maintains an overview of initiatives and promulgates good practice.

2 *Raising awareness and motivation to learn in practising professionals*

The Centre should aim to accelerate the normal processes through which professionals become aware of their learning needs and are motivated to learn. The programmes that it will seek to develop must therefore aim to bring awareness of genetics applications to the attention of a variety of professional groups, through both formal learning and in the processes of providing clinical care. Our workshops have told us very firmly that raising awareness and motivation should be based on current realities with tangible benefits for patients, rather than on future possibilities.

(a) *Formal programme*

The formal programme to raise awareness and to acquire the rudiments of genetic science is the easier one to envisage. This is based on the very substantial learning that professionals undertake as part of their commitment to keep up to date: attendances at meetings, conferences, ward rounds and their personal work in research, teaching and personal reading.

The Centre should seize every opportunity to report developments in genetics and to link them to everyday practice. It would, for example use its Steering Group and key contacts in professions and educational institutions to seek opportunities for genetics topics to be included at conferences or seminars and in journal articles. Such activities are labour intensive and would require:

- Opportunities to be sought through systematic scanning of events announcements
- An ability to provide links to potential speakers and, if necessary, support them in preparing work
- Capacity to spot opportunities, using science writers to place articles and to work with specialists to prepare drafts of articles on current developments and initiatives

Links to knowledge parks, the Public Health Genetics Unit, the Genetic Interest Group and others would assist in the information gathering and network infrastructure. The Centre should also, itself, put on a few events and workshops to raise awareness and educate on key topics and with key professional groups as identified by the Steering Group.

(b) *Learning needs arising from clinical practice*

The stimulation to learn within clinical practice is harder to reinforce on a systematic basis. A programme must be based on a very firm understanding of both the general evidence of what works in motivating professional learning and the particular evidence of effectiveness in genetics education. The general ways by which professionals become aware of learning needs through clinical practice include:

- Encounters with a clinical picture that is entirely unfamiliar
- Difficulties arising in clinical practice, through audit, during discussions or occasionally after a complaint
- Consideration of innovations in practice
- Knowledgeable patients
- Clinical mistakes

In addition, some professionals go through relatively formal process such as diary keeping, or reflection on clinical practice.

As the numbers of genetics applications increase, so professionals will become more aware of areas in which they lack expertise. At present, professionals still encounter these areas relatively infrequently; or the genetics aspects of services (for example, antenatal screening, or the care of patients with chronic diseases) are so embedded that the professional does not consider any special genetics expertise to be important. Often professionals are simply not aware of their own ignorance and incompetence. Thus ways of bringing genetics issues to their attention at critical times could be helpful.

It is recommended that the Centre investigates further ways of motivating professionals to learn about genetics through clinical practice. This might be through:

- Identifying those caring for particular groups of patients (e.g. a team looking after children with cystic fibrosis)
- Targeting those who have made particular referrals or requested particular tests (e.g. professionals referring patients with family history of cancer)
- Design of audits that directly tackle areas where there might be learning needs, for example, in provision of support for informed consent in antenatal genetic screening services

These would then be accompanied by bringing to their attention educational resources that might help them deal better with these areas.

A second possibility would be to develop a tool that could be used in the context of appraisal or the development of personal development plans. Such things might include a tool for self-assessment in genetics, which usually involves some sort of reflection on practice.

(c) *Promotion of the Genetics Education Programme*

Promoting developments in genetics should be underpinned by a process to actively promote its own work and the work of its network of partners through the publication of a newsletter, the development of a Centre website and a presence at conferences and other events.

We recommend that the Centre should pursue a programme to raise awareness of genetics amongst health professionals using formal opportunities, by building on needs arising through clinical practice and by promoting its own work.

3 *Pursuing formal opportunities to promote genetics education in health service provision*

(a) *Policy work*

All stakeholder groups mentioned the importance of including genetics among the priorities set by the Department of Health for the NHS. This was thought by many to be essential. The plethora of competing priorities within the NHS demand at least a modicum of political steer to allow managers and others an 'excuse' to give some attention to genetics in their organisations. The National Service Frameworks also present clear opportunities for raising awareness. The monitoring processes of the Commission for Health Audit and Inspection provide another route of influence. Other formal opportunities noted in the White Paper include the work of the National Horizon Scanning Centre, National Health Technology Assessment programme and the National Institute for Clinical Excellence. These initiatives would provide a forecasting mechanism and guidance on effective healthcare; and we strongly suggest to them that educational needs should always be taken into consideration in their deliberations.

The coordination of all these mechanisms and the bringing together of available expertise would require a huge amount of energy and could only be catalysed by a formal organisation such as a Centre for Genetics Education. This would need to work with other communities such as the Department of Health, the Public Health Genetics Unit, Knowledge Parks, and the Joint Committee on Medical Genetics to:

- Identify opportunities,
- Make contact and negotiate agreed input process
- Propose and organise suitable individuals to consider genetics education aspects of input alongside general genetics aspects
- Ensure content is developed and suitable recommendations made

(b) *Building education on service developments*

The many areas where genetics is becoming a factor in new services offer a formal opportunity to consider the educational needs of practitioners and the ways in which they could be met. They provide particularly useful opportunities for work-based inter-professional education. Some examples of expected or current developments are given in Box 3, overleaf.

Box 3 Service developments where genetics education may be an important factor in implementation

- *New national screening antenatal and neonatal genetic screening programmes for haemoglobinopathies, Down syndrome. Education is currently being commissioned in support of these programmes*
- *National Screening Programme pilot studies such as those on colorectal cancer*
- *The development of cancer networks. There are many opportunities to consider what competencies are required to ensure the programmes deal competently with hereditary and familial cancers*
- *The development and subsequent implementation of guidelines for those with a family history of breast cancer – currently under consideration by NICE*
- *The development and implementation of decision support software for people with a family history of breast and other cancers*
- *Development of coronary heart disease networks*
- *Pilot studies for the implementation of cascade screening for family members of people with familial hypercholesterolaemia*
- *Developments in pharmacogenetics*
- *The availability of over the counter genetic tests*
- *New genetic tests. The genetic testing network has developed a procedure for the evaluation of potential new genetic tests. The process of implementation could include a formal consideration and evaluation of the education requirement for those involved in offering the test and others in the care pathway.*
- *Enzyme replacement therapy*
- *The development of new clinical roles, such as general practitioners with special interests and nurse consultants in certain key areas such as cancer nursing, paediatric nursing, learning disabilities, haemoglobinopathies and diabetes. This could include a consideration of competencies in genetics.*

Source: Hinxton Workshop May 2003

Funds have been allocated in the White Paper to stimulate a wide range of initiatives in England that involve genetics development in the mainstream of NHS care with £2 million each to be invested in secondary and primary care; and it is of some concern to the developing educational strategy that health services in the devolved administrations have not made similar provision. Possibilities such as the setting up of joint clinics, or the development of referral or good practice guidelines are mentioned. It is recommended that the inclusion of consideration of an evaluation of the roles of the various health professionals and their needs for genetics education is an explicit requirement in any bid for funds. Once established, the Centre should maintain an overview of this aspect of the work with a view to helping to steer it, making appropriate links to a range of experts both in genetics and in education, and ensuring that materials developed and other learning arising from the work are made accessible within educational arenas.

One area where the development of professional education programmes has been commissioned concurrently with the planned implementation of new services is the development of the new antenatal and neonatal screening programmes for Down

syndrome and haemoglobinopathies (now known as the Sickle Cell and Thalassaemia Screening Programme). These programmes, which will cover both the necessary knowledge base and also skills in communication for informed consent, must be integrated with the developing Genetics Education Programme.

When new and existing genetic tests are evaluated within the Genetic Testing Network Framework, consideration should be given to the implications for education of professionals involved with these patients. This reflects the observation that, although most tests will be requested by a specialist geneticist, it is likely that the doctor with overall responsibility for clinical care will be from a different speciality and may not be fully knowledgeable about genetic aspects.

Further educational work might be linked to more local service developments. The opportunity might be used here to seek out developments over a wide range of providers, health professional groups, service areas and client groups in order to consider educational needs. Examples here might include a consideration of the needs for genetics education of professionals involved in care for children with learning disability, or throughout the patient pathway, from specialist to community care for patients with a chronic multi-system genetic disorder such as cystic fibrosis or Huntington's disease. Any of these groups could include a wide range of dietitians, physiotherapists, occupational therapists, speech therapists, and even extend outside the health service to social workers, those in the educational services and voluntary organisations, all of whom are part of the wider team.

Such work will require first an ability to spot opportunities and negotiate a 'genetics education' input; and second, the commitment of resources to provide the necessary adjunct to the developmental work. This would include specialist subject input, focus work with professionals to identify needs, development and evaluation of educational support programmes and subsequent rollout to other areas. This should run in parallel with the pilot or developmental work.

We recommend that the Centre should pursue formal opportunities to promote genetics education within the NHS by identifying, promoting and coordinating involvement in national policy work, and by promoting and contributing to the integration of genetics education as part of service developments.

4 *Promoting the development of leaders and facilitators with a special interest in genetics outside specialist services*

Although much learning takes place through personal study, there is also an important role for local groups or networks, department or practice training sessions, seminar series or journal clubs. Genetics must be built into these local processes, but to do this we need local experts and product champions, preferably fellow professionals from within that peer group, who have become sub-specialised and are able to extend their expertise in genetics from the firm stand-point of a thorough understanding of local professional needs. These individuals will be able to provide potent learning and, because it will be highly focussed and relevant, they will also be able to promote awareness and ownership for genetics and the genetics education programme outside the specialist centres. Finally, they will be able to give

confidence to local professionals in introducing genetics into their own practice by giving advice and assistance when queries arise.

The Centre should work to encourage and allow professionals in other specialties outside genetics to sub-specialise in relevant genetics aspects. Good examples already in place, but very limited in number, include:

- The establishment of a GP specialist registrar in genetics in West Midlands
- The establishment of a training posts specialising in public health genetics at the PHGU, the School of Health and Related Research SCHARR at Sheffield and in Cardiff
- The use of coordinators for the genetic screening programmes in antenatal care

The Programme should use its influence to ensure that these posts are extended systematically so that there are local specialists in each geographical region and professional group.

The implications of developing sub-specialists are substantial. For doctors, they will impinge upon the royal colleges and the regulations that exist on training specialists. The idea that there should be cardiologists, dermatologists, vascular surgeons or oncologists, for example, with an interest in genetics will require negotiation with the Colleges and may require some change in Calman regulations. Capacity will also be an issue in medical genetics departments which is where at least some of the training will take place.

In the White Paper the government made a commitment to the introduction of GPs with a special interest in genetics and will provide start-up funds for their establishment. In other areas where such practitioners are established, they have a remit for education and liaison and service development as well as providing more specialist clinical services. This pattern may be of use when considering roles for those in genetics, although the starting point, usually a group of GPs with specialist expertise, will be somewhat different in the case of genetics. Opportunities should also be sought to develop further nurses who would take a special interest in genetics within a given setting such as primary care, or within service areas such as paediatrics, cancer care, learning disabilities or haemoglobinopathies.

The Education Centre should support the educational roles of GPs and other practitioners with a designated special interest in genetics by bringing them together in a network that will coordinate their educational work and give them access to national expertise and educational resources.

We recommend that the Centre should promote the development of professionals with a special interest in genetics and support their educational role by bringing them together in assisting them in developing and accessing educational resources.

5 *Developing core competencies in genetics*

Each profession has its own share of medical knowledge and a set of professional competencies, which together allow it to fulfil its agreed set of roles within healthcare. Although they are based on an understanding of the same sets of scientific, clinical and social knowledge each profession will pick out a different subset. This individualisation of learning need continues into the various areas of specialist training, such as that of cancer nursing, primary care, dermatology or paediatric dietetics.

Our workshops supported the view that the educational needs of all these practitioners are each interpretations of genetics finely tuned to individual circumstances. Though there would be some possibility of overlap it would be an expert task to develop formal education for each group, which would need to include both theoretical and practical aspects. The motivation to learn and the acquisition of new knowledge and skills can only be achieved if the practitioner sees the relevance through his or her own work and has the chance to consolidate through practical application.

We recommend that the Centre should work with the various statutory and professional bodies and educational establishments to ensure that:

- Genetics education is embedded in the general education at undergraduate, professional, specialist and continuing professional development levels as appropriate for that profession
- Consideration is given to training and accrediting physicians, nurses and others with a special interest in genetics but outside the specialist field

All professions need to progressively develop core competencies or learning outcomes in genetics, specific to their profession and to the different levels and specialties. This should build on work already progressing and be systematic. Though it will be developed largely by those with an interest in genetics it is important that outlines become accepted by the relevant professional organisations and are mapped to and embedded in current curricula.

Formally, as curricula are reviewed the opportunities should be taken to include genetic aspects. On the other hand, as core competencies in genetics are developed for the various groups these should be pursued actively with the appropriate statutory and professional bodies to ensure that genetics is included.

Informally, most curricula are fairly general and so the degree to which genetics is included is open to wide interpretation. There are also possibilities of introducing topics on genetics into other areas. For example, the teaching of communication skills might be based on the topic of informed consent for an antenatal genetic screening programme. Thus, when good genetics education programmes are developed, they should be actively marketed in other areas of the curriculum.

For undergraduate medicine the essential core knowledge and skills have now been agreed⁵. The next step is to ensure that they are implemented in each medical school and that supporting resources, such as the development of more complex scenario-based teaching material is developed collectively and shared between schools.

For postgraduate medicine the work led by the Centre for Research in Medical and Dental Education at the University of Birmingham⁶ to develop an agreement on core knowledge and skills to be incorporated into higher specialist training should provide a foundation on which other specialties could build. This work is to develop an agreement on core knowledge, skills and attitudes to be incorporated into higher specialist training, initially for three specialties. We understand it is currently being extended, with funding from the West Midlands Deanery, to explore the needs of those in general practice training, thus supplementing preliminary work by the GP Virtual Genetics Group⁷. These outlines will then need to be promoted with providers of higher specialist training, included in formal curricula and in areas for examination.

For nurse education Dr Maggie Kirk in the Genomics Policy Unit, University of Glamorgan⁸ in partnership with the Cancer Genetics Service for Wales has led work to identify core competencies. This provides a very solid base on which to build more detailed curricula. These will include not only basic education for nurses, health visitors and midwives, but also areas of more specialist competence for nurses in speciality areas of haemoglobinopathies, cancer care, paediatrics, learning disability and primary care.

Teaching programmes for nurses must cover the entire range and be specifically designed to be appropriate to their existing science knowledge, to be used in a variety of teaching styles and have some emphasis on the social implications of genetics so as to appeal to their perceived emphasis on education for holistic care.

There will be a problem in identifying teaching faculty with sufficient breadth in genetics in each nursing school. Specific ways of overcoming this should be explored. In particular, when teaching programmes are developed they should include consideration of how teachers need to be trained and how they can be given appropriate support. This might include suggestions on where they can access

⁵ Haites, N et al. Teaching Medical Genetics to Undergraduate Medical Students. 2002 Document agreed by British Society for Human Genetics and Joint Committee on Medical Genetics available at [www.bshg.org.uk/Official%20Docs/UNDERG~1.doc]

⁶ Wakefield S et al. Genetics Education: Needs and Evaluation (GENE). First interim report. Centre for Research in Medical and Dental Education at the University of Birmingham. April 2003

⁷ Genetics in General Practice. A guide for VTS tutors. RCGP Virtual Genetics Group. Convener Rhydian Hapgood

⁸ Kirk M. Fit for practice in the genetics era: defining what nurses, midwives and health visitors should know and be able to do in relation to genetics. Genomics Policy Unit, University of Glamorgan. June 2003

specialist information, special material such as patient accounts or local experts who might be asked to participate.

Pharmacists have an extensive science base in their general training. The Royal College of Pharmacists of Great Britain is currently reviewing core competencies for the workforce⁹ and it is important that this explicitly includes genetics across a variety of fields. These include the underpinning knowledge base, where, as well as the molecular aspects of genetics, pharmacists need to understand about epidemiology and measurement of risk, inheritance and pharmacogenetics testing, public health aspects, consultation skills about communicating risk, and explicit aspects of ethics related to pharmacogenetics testing.

A special working group should be set up of geneticists and pharmacists with a special interest in genetics to identify and determine the core knowledge and competencies at undergraduate and postgraduate levels. It should then go on to develop core materials, and a course for pharmacists that could be provided in undergraduate education and professional training and through the standard CPD channel for practising pharmacists.

The curriculum for **dietitians** needs to be based on preliminary work to set out a realistic analysis of the future need and scope of genetics within the field. As well as integrating genetics into the curriculum, special modules on genetics could be developed for the pre-registration curriculum and MSc courses. There is currently an opportunity to get genetics into the Standards of Proficiency for Dietitians as these are in the process of being written for the Health Professions Council, which is the regulating body for health professional groups. Genetics already appears in the Benchmarking Statement for dietetics (QAA, 2001).

For **public health professionals** there is no formal agreement in the UK on core competencies in genetics for public health specialists but the areas covered by the course provided by PHGU for public health and health service managers provides an outline of the areas that should be covered¹⁰. Work should be undertaken to identify those competencies in public health genetics that would be appropriate for those completing higher specialist training in public health medicine.

The development of a cadre of public health specialists in public health genetics to work in each region is also important and an outline of the competencies required at this level would also provide a useful starting point.

The competencies for **health service managers** will vary according to the area of work for the manager. It is recommended that the objectives for the education of managers in the current programme described in the Genetics White Paper, commissioned by the Department of Health and undertaken by genetic testing

⁹ Competencies of the future pharmacy workforce. Phase I Report. Royal Pharmaceutical Society of Great Britain. April 2003

¹⁰ Course details available on www.phgu.org.uk/newsletter/course/course.html

network and PHGU should be kept under review. Evaluation of the workshops should be used as a basis for further refinement.

6 *Development of educational programmes*

There should be a rolling programme, facilitated by the Centre for Genetics Education to work with expert groups from the professions and current providers of education to develop detailed educational material for different professional groups, and specialties. The work would need to cover undergraduate and postgraduate education and CPD. These should follow a standard methodology that would include an agreed range of stakeholders: representatives from clinical practice; voluntary organisations; educationalists; experts in e-learning; geneticists; experts in such areas as communication, ethical, legal or social aspects, depending on the areas to be covered; and, most importantly, people who can represent the view of service users.

The developmental work already underway should be brought into the frame. It includes the piloting of a local workshop approach with supporting educational material and programme outline for the education of health service commissioners, and the development of educational programmes to support antenatal and neonatal genetic screening commissioned by the National Screening Committee.

Priorities for the programme should be agreed by a steering group and should broadly reflect the priorities described in this document. As well as developing genetics in basic education for each professional group, priority areas would include:

- Cancer genetics
- Genetics as a component of disease in other hospital specialties outside genetics
- Antenatal and neonatal genetic screening programmes
- Obstetrics (pre-implantation, antenatal and neonatal genetic testing)
- Pharmacogenetics
- Chronic disease care for people with genetic conditions
- The development and management of specialist genetics services
- The implications of genetics for public health practice

A very important element of all these programmes will be the need to develop trainers and teachers. In some specialties, such as nursing, this will be a programme in itself. In other areas, consideration of support to trainers through the development of resource packs or education events should be concurrent with the development of the educational resources.

We recommend that a rolling programme of educational resource development is implemented covering each professional group and all levels on an understanding of needs and priorities.

7 *Facilitation and sharing of resources*

Until now, most genetics teaching has been done by genetics experts, often from the regional genetics centres. We learned from our survey of those providing genetics education to undergraduate medical students that most develop their own materials, rarely with any recourse to electronic sources.

The impetus to develop jointly and share resources arises from three new aspects of genetics education:

- Genetics education will need to be delivered widely and there will not be enough expert teachers to support this. New teachers will be brought into the frame and they will need much more support by providing them with teaching materials.
- There is general acknowledgment that 'good resources' involve such things as guided clinical cases or direct patient experience that are more time-consuming, complex or costly to develop, or that require a wider range of expertise, including that of patients.
- The Internet already provides a wealth of educational material in genetics but finding a way round this can be complex and time-consuming.

Most teachers, in practice, do not access Internet resources, and will miss out on some excellent material. Their reasons vary but include not knowing what is available or how to access it; being unsure how authoritative it is; and the fact that it is not in a useable format or cannot be tailored to their own specific needs.

The medical genetics leads surveyed in our questionnaire indicated a desire to share the development of new resources and, in many cases, to share those they already had. This was endorsed by the stakeholder groups, where all professions recognised that readily accessible, high quality educational resources would be an asset that would save professional time and result in higher quality teaching.

One of the main functions of the Centre should therefore be to devise a system that will fulfil this sharing. This will rely heavily on electronically available materials. The need will be to add value to what is already on the Internet, whilst at the same time incorporating new developments and local work.

We would envisage this part of the Centre to be not simply a database of electronic teaching resources, but also an information service, that could point individuals to good materials and facilitate their access. A range of materials would ideally be maintained, labelled and cross-referenced so that teachers could 'pick and mix' according to their needs. It would include:

- Courses, learning modules or presentations aimed at particular professional groups, and covering the range of topics in the genetics spectrum from basic science to ethical, legal and social aspects
- Material relating direct patient experience – videoed, taped or written accounts
- Clinically based materials such as case histories, guided case examples, clinical problems
- Material related to different diseases or conditions

Basic primers of genetic science will also be needed. Cross-referencing material that would fit into a number of categories would be important and would have the additional value of being accessible to other teachers looking for more general material, for which there was a good genetics example (e.g. communication skills).

There is a danger that such a venture could be over ambitious. For example, it could not be expected to review all the educational material already present on the worldwide web. Some realistic aims would be:

- To ensure that material specifically developed and produced or reviewed within the Educational Programme and known to be quality assured, was made accessible to teachers, together with any formal evaluation of this material
- To keep a database of other UK material submitted by education providers, including purpose, audience, a note on content and any reviews of how it was used or evaluations
- To keep an overview of other educational material on the Internet, and maintain links with other teaching resources such as more general collections of patient material [www.dipex.org]
- To be able to give general advice on the content and quality of all these resources and how they can be used.

We recommend that the Centre should promote and facilitate the sharing and dissemination of educational resources through the development of a database and information service.

8 *The development of electronic resources to provide clinical support and information*

A programme to develop first class information, clinical support and educational resources for health professionals via the Internet is vital to ensure that professionals have the 'right information at the right time' and are enabled to provide a better service for patients and capitalise on learning opportunities as they arise in practice. This is a major programme that will require substantial funds, extensive professional clinical, educational and IT expertise and a high level of cooperation and collaboration between major UK centres. The Internet programme should be overseen strategically by the main Steering Group.

For most practitioners, the subconscious education that takes place as they solve problems in clinical practice is a powerful form of learning. Too often, at present, however, clinicians do not have the time or skills to gain access to the necessary guidance that can help them to manage their patients better, the support is not available or they are not confident that information is locally trustworthy.

It is recommended that current electronic resources, including developing NeLH sites, regional medical genetics centres and sites of professional organisations such as the British Society for Human Genetics (BSHG) website and the Royal College of General Practitioners (RCGP) virtual genetics website, be functionally linked and further developed. A resulting system could provide a central information portal giving reference information on genetics, guidelines, and links to websites of regional

genetics services, patient information, clinical decision support and sites with specialised information on particular genetic disorders. It could also be developed to provide access to specific educational materials in genetics.

The very variable nature and depth of regional genetics centres websites, as found in the review, could be enhanced by the development of a prototype that emphasises the information about local service provision and any national specialist expertise and relies on the central service for information that is generic, including links to other sites. The current situation in which regional websites are largely set up and maintained by enthusiasts will not be tenable across all organisations. Support should be given to encourage the development of regional websites and to assist in their maintenance, possibly through making available website expertise that is centrally coordinated.

Specialist websites should also be developed that can take the lead in various important disorders such as muscular dystrophies, cystic fibrosis etc. These could include guidelines or patient 'care cards'¹¹ that would give information on standards for patient pathways and care and would normally have been agreed by professional groups on a national or international basis. An example of such a specialist site is the Scottish Muscle Network. Further sites could be commissioned from existing specialist services.

The further development of computer assisted family history taking (see Box 8, page 62) and recording and tools that assist in risk assessment and link to management guidelines should be encouraged and those systems made available with appropriate educational support over the Internet.

We recommend that a major programme should be developed to provide access to authoritative information, clinical support and educational resources via the Internet. This should include further development and functional linking of NeLH genetics site, BSHG and regional medical genetics centre websites and provision of links to specific educational material.

4.5 Resources

For the purposes of considering future implementation of the Strategic Framework, we undertook further work to develop a view of the likely level of resource requirement for an education Centre and programme based on a set of assumptions that are used by way of illustrations and in order to obtain an approximate estimate.

¹¹ Available from the Scottish Muscle Network at www.gla.ac.uk/centres/muscle/dmcarecardgen.pdf

The following areas of activity would need to be funded:

- The running of the Centre, including the necessary staff, buildings and supporting infrastructure
- Providing administrative support to the Steering Group and its Chairman
- Providing input to the main strategic work and maintaining an overview of initiatives
- Commissioning the necessary educational development including consultancy from the appropriate subject experts
- Publishing educational materials in a variety of forms
- Commissioning educational work to run alongside service development
- Funding pilot development work on regional websites and enabling local setting up
- Running conferences, seminars, workshops
- Publishing a newsletter
- Running a database and providing an information service
- Providing a Centre website
- Providing administrative support to a network of educational facilitators

The resources required to run such a programme would be substantial. Success in the programme will be achieved only through the establishment of a significant presence and the completion of highly regarded work. We estimate that this would require an annual budget of about £700K for the Centre alone if it is to have the necessary impact. For a detailed breakdown of how this figure was reached see Appendix 3, which contains an illustration of staff and non-staff costs based on our experience in the Cambridge Genetics Knowledge Park.

The actual development of specific educational programmes would require additional resources to cover elements such as the use of expert groups, workshops, room-hire, critical reading, validation, assessment and evaluation. Following a general model devised in CGKP as part of a bid for a genetics education project commissioned by the National Screening Committee, we estimate that costs of this would be about £150K per programme. If four programmes were to be developed each year a further £600K would be required. This excludes the cost of eventual publication of materials. Such costs are difficult to estimate as it will be dependent on the number and types of materials required.

Further, this estimate covers the running of the Centre alone and does not cover budgets to pump-prime developments such as the use of regional coordinators or education facilitators, the development of practitioners with a special interest, or intensive programmes of 'training the trainer'. Such programmes would be labour intensive and would add substantially to the programme - perhaps in the order of £500K for regional coordinators alone.

The scope of the programme and its eventual utility to a wide range of stakeholders, including those interested in the applications of research, in the promotion of public health and health services, in the promotion of genetics applications within the private sector and in the development of health professionals, suggest that a

successful programme would best be achieved by a partnership of several organisations.

The Department of Health for England has already signalled its substantial commitment to genetics education and it is to be hoped that, following their reviews, the Scottish Executive, Welsh Assembly Government and the Department of Health, Social Services and Public Safety in Northern Ireland might also wish to become partners.

Beyond this, it is imperative that other organisations become involved. The Medical Research Council and other research councils, organisations and charities all have a keen interest in ensuring that the benefits of genetics research are realised in medical practice. The pharmaceutical industry has a large interest in the development and application of genetics, and also an ongoing commitment to support medical education. They should all be encouraged to underwrite their interest in genetics through a substantial commitment of resources.

We believe that it is only through the commitment of this level of resources by such a range of partners and over the forthcoming decade at least, that we can build a solid resource base for a continuing and substantial UK programme.

Section Two

The Evidence

5 An evolving public: social science themes related to genetics

5.1 Findings from the meeting with social scientists

Rare genetic disorders affect only a small proportion of the population, but it is becoming increasingly clear that genetic factors have an influence in all common illnesses. The impact of genetics as a determinant of health is therefore relevant to all of us, whether as individuals or health professionals. This explosion of interest in genetics is reflected in media coverage, in governmental initiatives and in new research and educational initiatives for both public and professional audiences.

What is less often considered is the 'environment' within which these developments are occurring, the importance of ethical, legal and public policy frameworks as well as opportunities for public engagement in the evolving science. On the other hand, society itself is evolving, and with it public concerns and shifts in values, attitudes and behaviours that will impact upon the ways in which applications in genetics are developed in health and healthcare.

The following themes, which may help provide a broad context for strategic planning of genetics education for health professionals, were identified through reflection and discussion with social scientists at the Centre for Family Research, University of Cambridge:

Nature and validity of 'knowledge' and 'expertise'. Increasingly the special understanding and insight that is gained by experience is considered a valid form of expertise (the 'expert patient').

Public access to information. An increasingly sophisticated, educated and proactive public claims its right to make informed choices.

Accountability of the medical profession to the public. Health professionals are challenged by the autonomous patient who demands involvement in decision making, accountability and transparency.

Evidence based practice. This is increasingly used to inform medical decisions but the professional must complement this with clinical judgment.

Personalised and individual needs. These will need to be balanced against public health and societal needs.

Lack of public enthusiasm for genetic testing. This, as well as worries about 'negative' test results, may prevent the opportunities for disease prediction and prevention being realised.

Attitudes towards health and illness. The tendency to medicalise conditions continues and in the future the way in which people make decisions about personal lifestyle in relation to genetic factors will be crucial. The right for patients and families not to know about susceptibility will be important.

Informed consent. The locus of responsibility for decision making about procedures and treatments that incur risk is shifting from companies, hospitals and clinicians to individuals.

Support for science and research. The public has become more critical of science, especially where ethical issues are involved.

The role of public participation. The public is being asked to take a consultative and audit function with regard to policy making and service delivery.

5.2 Main implications for education strategy

Integrating genetics into the clinical care for patients comes at a time when the relationship between patients and professionals is changing. Whereas previously the pattern was one where the patient was subservient to the clinician, a recipient of knowledge and advice about possible courses of action, in the 21st century we will see the emergence of the negotiated partnership. Patients will have resources and information to enable them to be actively involved in the planning and management of their own health and healthcare. Genetic diseases and concerns provide a perfect example of the importance of this new relationship.

As the science advances, the public and health professionals will need to incorporate genetics into their understanding of the factors that determine health and disease. The development of most common diseases is a complex process involving both multiple genetic factors and environmental factors - lifestyle choices such as diet, exercise and smoking. With a shifting paradigm of care from treatment to health promotion and disease prevention, professionals will need to be prepared as people seek advice about predisposition testing and lifestyle changes.

Most importantly, the need for real public involvement and accountability require that patients and the public be involved in the development of education strategy, of individual programmes and in the delivery of education.

Chapter 5 Policy points

Education in genetics should prepare professionals to provide their expertise in a more equal partnership where patients are also experts

Patients and the public should be involved in the development of the genetics education strategy, of individual programmes and in the delivery of education

6 The patients' perspective

6.1 The patient workshops

The purpose of involving patients in the consultation was to underscore their role as the ultimate stakeholders and to appeal for a patient-centred approach in designing education for health professionals in genetics. Defining the objectives of the education strategy in terms of improved health outcomes encouraged us to establish a baseline understanding of the current experience of patients and families diagnosed with genetic disorders and the role of many different kinds of professionals, in different settings, at different stages of illness – thereby delineating a broad scope for action. The 'patient perspective' formulated during the workshops suggests the education strategy should be informed by priority objectives that require the health professional to:

- Gain an informed understanding of genetic disease
- Ask: *Could this be genetic?*
- Communicate appropriately and sensitively with patients
- Reduce missed diagnosis and misdiagnosis
- Appreciate the impact of genetics information and diagnosis
- Develop standard protocols and 'joined up' systems for care
- Facilitate communication across disciplines and specialties
- Manage disease in partnership with the patient
- Provide sources of information and ongoing support
- Listen to our experience

These recommendations were presented to all stakeholder workshops and professionals were encouraged to discuss the educational interventions that would address these concerns, and to consider how patients might be involved in delivering the message of their own experience as part of an educational programme.

6.2 Main implications for strategy arising from workshops

Those affected by 'genetic diseases', where most of the current applications of genetics lie, will have new opportunities for diagnosis, testing and increasingly treatments. These are the inherited disorders, such as cystic fibrosis or haemophilia, caused by single gene defects. Genetic diseases have a long term and highly individual impact on the patient and family and these people will seek personal help and support. The new relationships will value patients as experts, recognising that only those experiencing the condition for themselves will have a full understanding of the social consequences of the disease in their own circumstances and be able to weigh up the risks and benefits of preventive or treatment options. This is a further paradigm shift that health professionals must recognise.

Furthermore, a widening array of genetic conditions is now being recognised. Altogether there are around 10,000 single gene disorders. Genetic testing is

currently available for about 200 of them but this number is set to rise over the next few years.

There is also an increasing level of detail on how family history might indicate susceptibility to disease, what can be tested for, how results need to be interpreted, and options for prevention or treatment. This means that health professionals cannot hope to keep abreast of the facts. Equally, patients with concern about particular conditions have great opportunities to access information on the Internet and are able to become very well informed about their own condition. Health professionals need to learn to value this different balance of information and use it to greatest effect, by guiding patients to relevant information and then bringing the professional element to aid decision making. They need the skills to search out relevant information for particular patients, and to be able to communicate and interpret it with them.

The patient with genetic disease has particular needs from health professionals. In many ways, these do not differ from the needs of other patients with rare and chronic diseases. The often protracted time before a diagnosis is made, the search for a specialist who understands the many aspects of the disease, the need to coordinate the input of the many different health professionals involved in care - often different system specialists - are all common features. Similarly, patients and carers note the difficulty of taking in a lot of new information at once and the need to spend time, usually on several occasions, to fully understand the nature of the disease.

Commonly, we heard that the patient with a rare genetic condition is virtually self-taught about the disease. Once given a diagnosis, many embark on a worldwide search for information and to find fellow-sufferers. The support and learning achieved through sharing experiences of living with the disease can be a huge relief and should not be underestimated. Indeed, the spirit of partnership with patients would suggest that they should be enabled to look for this support if they wish, and any resulting information and experience should be valued by the health professional.

The extra facet of genetics is that genetic conditions may have been inherited from, and can be passed on to, family members. This adds a whole new dimension of concerns, such as those over reproductive decision making, feelings of guilt, opportunities to pass on advice about prevention options, to which the health professional must respond. Where the patient is not in contact with a geneticist, or perhaps where this is some time in the past, the responsibility lies with the health professional responsible for care and others in day-to-day contact to pick up concerns or new circumstances and give advice or identify times when they might need more specialist support. An example was given in the workshops of how children with a genetic condition would need genetics advice for themselves when they become adolescents and start to consider possibilities of having their own family.

Chapter 6 Policy points

The education of health professionals in genetics must be centred on the needs of patients

The public and patients with genetic disease should be involved in the development of educational programmes

Real life accounts can have a high impact in education

Ways in which to include first-hand accounts should be promoted

Guidance, when using first-hand accounts, on the necessary safeguards for patients, carers and learners should be developed in the area of genetics

7 The problem of awareness and motivation

Every workshop raised the obstacle of lack of awareness about the importance of genetics and hence the difficulty in motivating professionals to learn.

7.1 Findings from the professional workshops

Promoting the desire and need to learn about genetics is fundamental to this strategy but the stakeholder groups all told us that the ground on which the seeds of learning might be sown is still relatively infertile. Whereas professional bodies and leaders of the professions are beginning to acknowledge the importance of genetics, and a few enthusiasts have foreseen the implications, opportunities and potential pitfalls for clinical practice, individual practitioners are, for the most part, still quite ignorant and unaware of the need to learn.

We learnt from managers that there is much scepticism fuelled partly by the perception that the potential of genetics is being exaggerated. The evidence for most of the clinicians was that genetics is not making a huge impact across the breadth of clinical medicine. The health problems and service questions with which they are concerned in their day to day work do not lead them to think that understanding genetics better will greatly improve their practice, or solve any problems. When faced with so many other clinical and health service problems, genetics does not appear above the horizon.

In fact, though conditions are individually rare, the patients with genetic disease can make up a substantial proportion of care provided. For example, 30 percent of all childhood admissions and between 40-50 percent of childhood deaths are due to genetic disorders or congenital abnormalities¹². However, we were told in the workshops that professionals looking after them do not consider it important to understand about the underlying genetics. This stems from a failure to recognise that there are different dimensions to genetic disease that might lead to further health needs, such as decision making about reproduction (see Box 4).

Box 4 Clinical examples from workshops

A specialist looking after a patient with neurofibromatosis did not recognise the implications for reproductive decision making when she was pregnant

Paediatric dietitians “did not see the relevance of them having a more general understanding of the genetic aspects [of cystic fibrosis]” when interacting with families.

¹² Mueller RF, Young ID. The history and impact of genetics in medicine. *Emery's Elements of Medical Genetics*, 3-10. London: Churchill Livingstone, 2001

Nurses may perpetuate their lack of awareness about genetics because they have dismissed it as 'scientific' and 'difficult' and not relevant to their central role of providing holistic care to patients and their families. Yet again, they can be motivated when the implications of the genetic nature of conditions and the wide ranging problems which can be produced for family relationships and future decision making are described in more personal terms.

Pharmacists, we were told, had a low level of knowledge and "did not know what technology existed, nor understand what was likely to happen". Nor had they given much thought to ethical, legal or social aspects, even where it might impinge on their practice.

This lack of awareness and motivation was rooted in the fact that current clinical utility is not widely accepted. Even where there are examples of current clinical utility these appear to individual clinicians as being small and rare. The mediocre service will rarely be exposed without incentive to improve the quality of practice. Some examples are given in Box 5.

Box 5 Examples situations in which there may be a lack of high quality service

Example 1

A pregnant woman going for Down Syndrome screening will not often question whether she has really given 'informed consent'- that is consent helped by a primary care professional who really enabled her to understand the option, benefits and risks and the pathways through the full screening programme. Decision making through this programme is incremental for patients. In the extreme it may start with the blood test, then what to do about a positive result, decision to have an amniocentesis and finally consideration about termination of pregnancy. The trauma of having to deal with an unexpected amniocentesis result, not necessarily a Down syndrome result, but possibly a different chromosomal disorder or even an unusual translocation of unknown significance will probably completely mask the fact that she may not have understood about the screening in the first place. However, quality standards for a screening programme will rightly demand evidence of initial informed consent.

Example 2

Each primary care practitioner may only very occasionally see a patient concerned about a family history of cancer. Failure to deal with each one competently will not apparently have a great effect. On the one hand, false reassurance and dismissing a positive family history might result in a failure to prevent disease, but not for a few years. On the other, simply referring them all for specialist genetics advice has the potential to overwhelm specialist genetics services.

Example 3

The patient with genetic disease is fairly resigned to being a rarity who, essentially, has to be self-educated to manage the condition and gain appropriate advice, and tends to go straight to the specialist for help. The small numbers of patients mean that this group does not constitute a loud voice calling for change in each practice, but individuals would be helped greatly if some of the professionals around them were aware of the family nature of this condition and recognised times when they needed further genetics advice.

As we found out during the course of the stakeholder workshops, when they do take the time to understand a bit more about genetics and to hear of the implications, professionals from each area can see the relevance and excitement of the subject. They then appreciate its potential to revolutionise their practice, resulting in real health benefits for their patients if appropriately applied, and awkward problems if not dealt with properly.

7.2 Organisational factors for raising awareness

What will be the main organisational factors for raising awareness to ensure that an educational programme is put in place? The question of how to raise awareness and increase motivation arose in each stakeholder workshop and was again raised formally in one of the Hinxton sessions.

(a) *"Getting it into the curriculum and motivating people to learn it"*

During formal education the teachers will be the prime source of awareness raising through their ability to present genetics as being part of the science that underpins medical knowledge and clinical practice. This will not be as a stand-alone part of the syllabus, and should be interwoven into teaching that is systems based, or problem orientated. There are genetics aspects to almost any question that might arise in clinical medicine.

Undoubtedly, the most important aspect that motivates students to learn is knowing what will be examined. If genetics is to be taken seriously by students it must routinely feature in examination questions or assessments. Whilst part of this will be to ensure that genetics parts of the syllabus are examined, as genetics becomes more mainstream it should become expected that consideration of a question would not be complete unless genetics aspects had been covered.

(b) *Inclusion in government priorities*

The official underpinning in the government White Paper of the importance of genetics becoming mainstream in NHS services will provide an important impetus and much would be achieved by ensuring that priority for genetics was included within such national initiatives as National Service Frameworks.

(c) *A formal programme to raise awareness*

Dietitians and pharmacists were in favour of a formal programme to raise awareness. For both, the starting point would be a formal analysis of the likely future scope of genetics and its scientific and professional implications. This would need to result in a position paper and strategic plan. There should then be a conscious programme of activities to raise awareness for the profession, including professional organisation activities, conferences and seminars, newsletters, fact sheets and articles in the specialist press.

(d) *Using patients as a resource to raise awareness and motivate*

Clinicians' awareness and interest is stimulated at first-hand by the problems confronting them each day in surgery or other clinical practice. They want to respond to the needs of their patients. If we can find ways of making patients more

demanding in these areas we might be able to stimulate professionals to seek further education. One example was given to us of the way in which a specialist information resource about myotonic dystrophies, the care card, was made available directly to the public via the Scottish Muscle Network¹³ website. In this way, it could empower patients in their interactions with health services by helping them to understand expected standards for care and to encourage their own professionals to attend educational sessions. Similar models could be used for other specialties and in other geographical areas.

Chapter 7 Policy Points

Awareness of genetics will be most effectively raised in the context of current clinical utility

Opportunities must be sought to ensure that competence in genetics is a key component of high quality services

Raising awareness of genetics should be a key objective of the strategy by:

- ***Ensuring inclusion and assessment in educational programmes***
- ***Promoting it in government standards for health services***
- ***Using patients to motivate professionals***

There should be a formal awareness raising programme as part of the strategy

¹³ Available at www.gla.ac.uk/centres/muscle/dmcarecardgen.pdf

8 Priority areas for genetics education

8.1 Priority areas expressed by health professionals

The government has signalled its intention that genetics will become more mainstream in NHS services. It will move out from the small speciality of genetics into every other hospital specialty and into primary care. The stakeholder workshops were all very aware that genetics was becoming more prominent outside the specialist genetics service. The main issues discussed and described as areas of concern are described below.

(a) *Primary care*

As the applications of genetics become more diverse health professionals in primary care will have a frontline role in incorporating this into clinical practice. With their responsibilities to support families through the whole pathway of care from prevention to continuing care they will need to be able to respond appropriately by:

- Dealing with patients' concerns about possible genetic disease
- Assessing and interpreting risk and communicating this to patients and their families to facilitate informed choice
- Genetic screening
- Referring to specialist services
- Supporting those with chronic genetic disease

Cancer genetics is an area that has already produced significant impact in primary care. Guidelines have been developed that allow at least some of the concerns of patients to be addressed at this level. For example, the West Midlands Family Cancer Strategy publishes guidelines on its website that assist primary care professionals in managing people with a family history of breast, ovarian or colorectal cancer: <http://www.bwhct.nhs.uk/wmfacs/index.htm>]. Through gathering basic details on family history they can ascertain who is at risk and reassure patients at lower risk. Those with lower risk can then be totally managed and advised in primary care and only those who might benefit are referred to specialist genetics department for further assessment and possible testing. The whole integrated programme does imply, however, that those in primary care have access to the guidelines, are able to carry out the simple family history assessment, are confident to discuss risk, and can access and provide the relevant advice for those in low risk categories.

This is an example which builds on a thorough epidemiological understanding of disease risk related to family history, as well as the role of relevant mutations such as the BRCA1/2 mutations in breast and ovarian cancer and finally, an assessment of the evidence for prevention for people in the different risk categories. Though the scientific and clinical applications have been much advanced in cancer genetics, it is likely that further opportunities will arise in assessing risk for people with family histories of other common diseases such as coronary heart disease or diabetes, with

the aim of identifying high-risk subsets where there may be significant options for prevention.

Further into the future as genetic testing technology gets simpler and cheaper and we have a better understanding of the relationship between genotypic variation and disease primary care, professionals may need to undertake the following:

- Genetic testing for susceptibility to common diseases such as coronary heart disease or asthma in order to give personalised advice on lifestyle or prevention options
- Genetic testing to support or refine diagnosis or to inform the choice of treatment. Here, the GP may be involved in a new set of interactions with pharmacists when it is decided who will undertake responsibility for the testing and final choice of prescription.

(b) *Hospital specialties outside genetics*

Most patients with genetic diseases are cared for by the appropriate system specialist, such as a dermatologist, neurologist, gastroenterologist or haematologist. They do not all get referred to specialist genetics services and, if they do, this can be a short consultant episode without any follow-up or further contact. It follows that the current specialist, whether doctor or, increasingly, specialist nurse, is responsible for understanding and identifying important genetic aspects of diseases within their specialty, including the implications for how they might advise their patients and other family members, the indications for referral to medical geneticists and for genetic testing. These opportunities can be missed if the specialist is not knowledgeable and skilled in caring for people with genetic diseases. Examples of this include identifying patients with familial hypercholesterolaemia amongst those with coronary heart disease or high lipid levels, finding patients with maturity onset diabetes of the young, or patients with inherited cancer syndromes.

(c) *Obstetrics and antenatal care*

In obstetrics services there is a real need for clinicians, including obstetricians and midwives to understand the opportunities and implications for antenatal testing and pre-implantation diagnosis. This means that they must have knowledge and skills in taking family history so that they can detect and respond to concerns about possible genetic disease. They must be able to take an initial family history and be able to discuss, at least initially, what the implications might be, what tests can be done, their laboratory and clinical validity and the options available for patients and their partners.

(d) *Antenatal screening*

Particular issues arise in the antenatal screening programmes where clinicians have to be knowledgeable and communicate adequately about tests to ensure informed consent. This would apply to Down syndrome screening and haemoglobinopathy screening, both the subject of new national screening programmes, and also to ultrasound screening. Screening implies that a test has been offered proactively

rather than that patients have sought services because of concerns. The health service thus shoulders special responsibilities to minimise any harm that might arise from the tests, and especially false positives and false negatives. All doctors and nurses undertaking antenatal care need some competency in genetics and risk communication for informed consent. This has been recognised as a particular need and is the subject of education programmes being commissioned by the National Screening Committee.

(e) Pharmacy practice

Pharmacists will be involved in pharmacogenetics where genetic testing may enable drug choice and dosage to be tailored to the individual patient with the aim of maximising response and minimising side effects. However, they also recognise an important role for themselves as, frequently, the first professional point of contact for patients with health concerns. In both these areas they have an important role in helping to shape a patient's understanding of disease and eventual response to treatment:

"No matter how good and potent the medication, the ways in which the patient uses it and their understanding and belief about the disease has a fundamental influence on the process of restoration of health or amelioration of ill-health."

Pharmacists workshop 28th March 2003

(f) Dietitians

For dietitians the current clinical relevance of genetics is in relation to risk of diseases such as diabetes or phenylketonuria (PKU). They also have substantial involvement in providing care for people with single gene disorders such as cystic fibrosis or Huntington's disease. As they are often in contact with families over long periods of time, they may well be asked questions or become aware of new circumstances, such as further family members with reproductive concerns, and need to be able to respond appropriately.

New research will present opportunities for more accurate assessment of individual risk and refining dietary management according to an individual's genetic profile.

Dietitians might be involved in giving advice about commercially available gene tests that claim to be able to provide individual profiles of risk based on a series of gene tests and linking this to personalised advice on various aspects of lifestyle.

(g) Health service managers

The role of health service managers is fundamental in ensuring the gradual expansion of genetics in service developments, and of a competent workforce to deliver it. As announced in the White Paper, this will include the development of specialist genetics services, of laboratory services for genetic testing, of workforce development and of implementing genetics within mainstream services, both secondary and primary care.

At local level they will each need to be knowledgeable about genetics as discussions on genetic aspects of disease and treatment become part of the normal debates in the processes of planning, commissioning and providing services. The place of molecular genetics laboratories in discussions about pathology modernisation will also require a basic understanding of genetics issues. As for other health technologies, if they are to make judgements and prioritise about the relative merits of genetics and other services they need to have a sufficient grasp of the basic science, clinical possibilities, ethical, legal and social aspects to understand and be able to speak authoritatively.

As genetics is already incorporated into a lot of services, such as antenatal care, managers must recognise the specific learning needs of their professional staff and ensure that education is commissioned accordingly and staff encouraged and enabled to attend.

Some priority areas mentioned by managers in the stakeholder workshops are given in Box 6.

Box 6 Priority Areas Mentioned by Managers

- *Antenatal and neonatal genetic screening, antenatal testing, family history of cancer, disease prevention and the integrated teams around the management of chronic disease with social care partners*
- *Maintaining quality and ensuring developments in the specialist genetics services*
- *Identifying and managing patients with single gene disorders within medical and surgical specialities*
- *Advising on the possibilities of prenatal and pre-implantation testing within the speciality of obstetrics.*
- *Developing integrated clinical networks, such as the cancer and coronary heart disease networks, which ensure that primary, secondary and tertiary care teams are working together effectively*
- *Ensuring competency of the workforce*
- *Overall pattern of service provision*

(h) Public health and health promotion

Public health specialists and specialists in health promotion must begin to incorporate genetics, as one of the fundamental determinants of health and disease, into their work on disease causation, prevention, health promotion and the development of health services. In the longer term, as genetics becomes more evident as an important component for many common diseases, public health and health promotion specialists will need to consider ways in which these factors might help target health promotion and disease prevention interventions. The role of genetic factors in the determinants of health and how these interact with the more conventional environmental, social or behavioural factors in the genesis of common diseases will need to be part of the core understanding of public health professionals. Their epidemiological base will have to be supplemented with an appreciation of the

techniques of genetic epidemiology, and of the pitfalls and difficulties of appraising this aspect of the scientific literature .

In the immediate term, they will be concerned, mainly in their commissioning roles, with the ways in which genetics is impacting on current practice. The public health roles of needs assessment and evaluation of effectiveness will increasingly demand an understanding of genetics. Although much of the detailed work will be done at a national level, in the immediate future at least, primary care trusts are currently being encouraged to develop collaborative commissioning arrangements for specialised services, an area in which public health specialists will become involved. In the longer term, when prevention or treatments are based on genetic considerations or the results of genetic tests, public health specialists will need to understand enough to weigh up evidence and make recommendations.

The ethical, legal and social context of disease has always been of importance and interest to the public health professional, but nowhere will this be as important as in the integration of genetic science into research and clinical practice. Issues such as data protection, the law on human tissue, and testing and screening have all been subject to increased scrutiny as a consequence of the genetic revolution. A basic understanding of these issues will not be a peripheral matter but a core requirement for the modern public health practitioner, even in areas such as information systems or risk assessment that are not directly or overtly genetic in substance.

Chapter 8 Policy points

All professional groups have priority areas where immediate education in genetics is required:

- ***Cancer genetics***
- ***Genetics as a component of disease in other hospital specialties***
- ***Obstetrics***
- ***Antenatal and neonatal genetic screening***
- ***Pharmacogenetics***
- ***The multi-disciplinary team caring for people with genetic disease***
- ***The development and management of specialist genetics services and genetics within mainstream services***
- ***The implications of genetics for public health practice***

9 Current opportunities to promote genetics competence in the workforce

9.1 Introduction

The publication of the White Paper on Genetics has given a significant boost to genetics, enabling it to start to be recognised as one of the priority areas for development. This in itself will not be sufficient to ensure that genetics gets included but it will allow it to be one of the areas in which organisations choose to invest time and resources. This is the sort of helpful steer that was envisaged by health service managers:

Whilst some steer from above that places genetics within the context of some sort of priority would be helpful this will be insufficient to appear very prominently above the parapet of other competing priorities.

Managers stakeholder workshop 30 April 2003

Stakeholder groups discussed what sort of opportunities were available or could be used to develop genetics. Important NHS programmes such as those around evidence, guidelines and standards, the development of the workforce, new arrangements for regulation of medical education and training and developments in the range of more specialist services to be offered in primary care, all offer great opportunities for building in genetics.

9.2 Genetics in National Service Frameworks

National Service Frameworks (NSFs) were established to improve services through setting national standards to drive up quality and tackle existing variations in care. NSFs have already been published for mental health, coronary heart disease, older people's services and diabetes. The next phase of NSFs will cover children's services, renal services and long-term conditions. Within the field of cancer services the Calman-Hine Report¹⁴ and the NHS Cancer Plan¹⁵ have similar status.

The stakeholder groups recommended that genetics should systematically be included in the development of NSFs. This means that for each there will be an opportunity to actively consider the genetic aspects of the condition or client groups in question. This would require the involvement of experts in genetics and public health genetics. Consideration should be given to disease, or subsets of disease, where family history or genetic tests can identify individuals or family members who would benefit from some form of preventive action. This would logically lead, when

¹⁴ Department of Health, Welsh Office. A policy framework for commissioning cancer services. A report by the Expert Advisory Group on Cancer to the Chief Medical Officers of England and Wales, 1995

¹⁵ Department of Health. The NHS Cancer Plan. A plan for investment. A plan for reform. 2000

standards are set, to the inclusion of some level of competency amongst health professionals to realise these benefits.

Box 7 Influencing National Service Framework

The government is developing a National Service Framework (NSF) for children, young people and maternity services. The first part, Standard for Hospital Services¹⁶, has now been published alongside an 'emerging findings' consultation document setting out the areas in which work is being taken forward by External Working Groups, including: maternity services, the health of all children, mental health and psychological well-being, caring for children in special circumstances and disabled children, and the use of medicines in children.

The PHGU made a submission to the Ill Child External Working Group highlighting the substantial proportion of genetic disease in this group and the importance that the wider support team of doctors, nurses, health visitors, physiotherapists and others have some understanding of genetics if they are to work holistically with these families.

9.3 The NHS as an educating organisation

Most stakeholder groups recognised the importance of the developing NHS University as a potent force for the future education of the NHS workforce. As part of the modernisation of the health service, it will make training and development accessible throughout the NHS, focusing on staff development that will bring benefits through better patient care. Although the details of what it would do were still unclear, it would clearly need to be involved in any strategy for the genetics education of health professionals.

The NHS University will work in partnership with organisations such as the Open University, *learndirect* [www.learndirect.co.uk] and UK e-Universities [www.ukeu.com] to deliver learning to all NHS Staff. It is planned that people will learn in a variety of ways including at work, in tutorials, through the Internet, and face-to-face. The NHSU will design some of its own programmes but will also work in collaboration with universities and organisations through which it will commission training. The first students will be enrolled in Autumn 2003.

In its ability to assess learning need, develop or commission courses and market them in various packages to NHS professionals the NHSU is set to become a powerful force in education across the NHS. Though its priorities at present are fairly generic, (communication skills, first contact, health informatics, educator support, management skills) these same mechanisms could be used to develop genetics courses and market them across the NHS.

¹⁶ Getting the right start: National Service Framework for Children Standard for Hospital Services. Department of Health, April 2003

9.4 The Postgraduate Medical Education and Training Board

Stakeholder groups concerned with postgraduate medical education and primary care mentioned the importance of new structures for regulating medical education.

In October 2003 a new Postgraduate Medical Education and Training Board¹⁷ will be established which will have a remit for all postgraduate medical education and training, bringing together training for hospital specialities and primary care. The new body is intended to improve the supervision of postgraduate medical education, raise standards and provide quality assurance of education working with royal colleges and faculties. As the various specialties review their curricula within these new structure there will be opportunities to influence training with the aim of integrating genetics into education across the curriculum.

9.5 Development in primary care of practitioners with a special interest

The development in primary care services of practitioners with a special interest is one of the innovative developments promoted under the NHS Plan to improve access to high quality care for patients^{18,19}. The White Paper on Genetics has also mentioned the need to train a small cadre of general practitioners with an interest in genetics.

These primary care specialists will provide localised services in familiar surroundings. They will improve the management of the workload between primary and secondary care and the quality of referrals to consultants. By so doing they will improve their own skills and enhance career opportunities. All of this can be applied to genetics.

Both GPS and nurse practitioners are included in this development. For GPs, the role would include direct clinical care: receiving referrals from colleagues within primary care; education and liaison, both in support of clinical practice (advice over the telephone) and the provision of more formal support and training for PCT members; and a service development or leadership role within a network of care. This would include developing care pathways, links with secondary care, local guidance, and building skills with other professional groups locally such as pharmacists and nurses.

A similar initiative is available to nurses for the development of practitioners with a special interest. This recognises and capitalises on the skills that practitioners have developed above and beyond the competencies gained from initial training. The specialist nurse might work across a number of practices, or outreaching from a

¹⁷ The Postgraduate Medical Education and Training Board. Statement on Policy. Department of Health, July 2002

¹⁸ Implementing a scheme for general practitioners with special interests, Department of Health and Royal College of General Practitioners, April 2002.

¹⁹ Implementing a scheme for nurses with special interest in primary care. Liberating the talents. Department of Health, April 2003.

hospital or community trust. Examples of expertise used in this way include specialists in respiratory disease, colorectal care, the delivery of intravenous therapy in a community setting, sexual health diagnosis and treatment, heart failure, epilepsy, mental health, pain management in older people, children's continence. We already have examples of nurses trained in genetics counselling providing outreach in primary care and this would be a good model to expand.

9.6 Personal development, appraisal and revalidation

There are new opportunities to put genetics higher up the list of priorities for health professionals through the newly introduced, and more formal, systems of personal development, appraisal and revalidation.

Appraisal for doctors began in 2001 and now includes consultants, non-consultant career grades and GP principals²⁰. For doctors, in April 2003 the General Medical Council put in place changes whereby from January 2005 doctors will require an up-to-date licence to practise renewable every five years through a revalidation process²¹. Doctors will usually show evidence of good practice through their annual appraisal process. This will provide a regular and structured system for recording progress and, in particular, identifying development needs through a personal development plan which will support them in achieving revalidation.

Work on appraisal and personal development plans for other NHS Staff will be taken forward through the Knowledge and Skills Framework²². This will be used to describe the knowledge and skills a person needs to be effective in a particular post and using a review process to identify areas for development.

Chapter 9 Policy Points

The relevance of genetics should be systematically considered in each National Service Framework by the inclusion of specialists in genetics and public health genetics in the working groups. Standards for care should ensure that health professionals are competent to deal with genetic aspects of disease

A genetics education programme will require a thorough understanding and close cooperation with emerging NHS educational structures including those involved in NHSU, PMETB, the developments of Practitioners with Special Interests, and systems of Personal Development, Appraisal and Revalidation

²⁰ Good Medical Practice. General Medical Council, 2001

²¹ A licence to practice and revalidation. General Medical Council, April 2003

²² The NHS knowledge and skills framework and related development review. Department of Health, March 2003.

10 What do professionals need to know?

Information from stakeholder groups

10.1 Introduction

We used the stakeholder workshops to get some indications for the main topic areas and types of learning that would be favoured by the different groups. These revealed considerable areas of overlap, but a sometimes surprisingly different emphasis from the various professions. They can tell us how to focus programmes on the areas that professionals will find most useful for clinical practice.

Stakeholder groups identified topics related to genetic science, clinical and practical skills, clinical and practical knowledge, knowledge and skills that would support their development in genetics, and ethical, legal and social aspects. The topics mentioned by all the groups were rationalised as far as possible, a final list generated and priorities for each group assigned by a simple ranking process. This rated the top third of priorities generated by each group as three stars, the middle third as two stars and the lowest third, one star. For further details of method see Appendix 2.

10.2 Areas of concordance between groups

The areas where there was almost unanimous agreement on need were:

- Increasing understanding of the basic molecular biology of genetics, or at least enough to become 'genetically literate'
- Communication about risk and facilitating informed choice
- Ethical, legal and social aspects of genetics
- Taking a family history and being able to take appropriate action
- How to get further information

10.3 Topics prioritised by stakeholder groups

(a) *Molecular biology*

All groups except primary care professionals and managers rated very highly their need for some understanding of molecular biology (see Table 1). Whilst managers did not think they needed to have a deep understanding of the science, they did think it was important to have some understanding of the terminology "enough to ask sensible questions and understand the answers". Primary care professionals did not think they needed to understand much about molecular biology, or DNA, but would need to understand about inheritance. Pharmacists and managers thought they needed to understand more about the role of genetics in health and disease. Interestingly, although pharmacogenetics was mentioned by a number of groups, only pharmacists and dietitians thought that it was an important teaching topic.

Table 1 Priority given to topics in molecular biology

Topics	Postgraduate medicine	Nursing	Dietitians	Pharmacists	Primary care	Managers
Molecular biology	***	***	***	***		
Genetics literacy						***
Inheritance					***	
Single gene	*		**			
Multifactorial			**			
Gene-gene/environment interaction			***			
Recent advances/future of genetics						*
Role of genetics in health and disease				**		**
Pharmacogenetics			*	***		
Developmental biology						
Genetic risk						
Development of new medicines						

(b) Clinical and practical skills and support

The roles currently undertaken by professionals reinforced their perceived need to learn more of the relevant clinical and practical skills – see Table 2. All practising professionals gave the highest priority to help with communication and being able to discuss risk with patients and helping them to make informed choices. Doctors and nurses in hospital medicine and primary care settings all gave priority to being able to take a family history and to interpret this and act upon it, referring where necessary.

Doctors also recognised that they will need continuing help with practical aspects such as the taking and recording of family histories, and the assessment of risk from these family histories in order to give appropriate advice to patients, especially those for whom specialist referral is not necessary and who can be reassured and managed totally in the primary care sector. There would be particular interest in the further development of tools that are available to assist in taking a family history and to assess risk such as that developed in Cambridge for breast and ovarian cancer (see box below). These were welcomed and should be made available via the Internet.

Box 8 Genetic Risk Assessment on the Internet and Decision Support (GRAIDS)

GRAIDS is a web-based computer program that supports the taking and recording of family history, advises the clinician about management and presents individualised risk information. Currently an optimisation trial is underway in primary care whereby a lead clinician from each general practice receives training in familial cancer and the use of GRAIDS and manages all patients in the practice with concerns about a family history of cancer.

The study will examine outcomes including number and appropriateness of referrals and patients' informed decision making.

*Department of Public Health and Primary Care, University of Cambridge
www.iph.cam.ac.uk/review/emery.pdf*

As a lower priority, some doctors and nurses felt they would like to understand more about the processes of counselling to be able to advise patients what to expect, and a small number wished to learn counselling skills. Also mentioned but given a lower priority rating was learning to listen and learn from client's experience. This was particularly valued by nurses, but was also felt to be important for managers and doctors. Though many had mentioned the importance of understanding other's roles, and the ways in which collaborative working might be beneficial for patients with genetic conditions, only pharmacists gave this a high priority. This was interesting and probably reflects pharmacists' perception that they are more peripheral at present in the multi-disciplinary team, but potentially have a lot to offer.

Table 2 Priority given to clinical and practical skills

Topics	Postgraduate medicine	Nursing	Dietitians	Pharmacists	Primary care	Managers
Taking a family history	***	**			***	
Interpreting and acting on family history and knowing when to refer	***	**			**	
Communication - about risk and facilitating informed choice	***	***	***	***	***	
Counseling	**	**				
Listening and learning from clients' experience	*	**			*	*
Collaborative working/the multi-disciplinary team		*		***		*
Assessing risk to patient			*			

(c) *Clinical and practical knowledge related to genetics*

We separated knowledge from skills because professionals indicated that there were some areas where they would need to increase their personal knowledge (see Table 3). Nurses and doctors in primary care and hospital medicine indicated that they would like to know about the conditions particularly relevant to their practice. For those in primary care these would include the genetics of common cancers, and the hereditary factors in common conditions such as diabetes and coronary heart disease.

Although four groups mentioned screening programmes, and this was much talked about during the workshops, only managers actually gave it a high priority. This probably reflects their perception that these programmes would be under a spotlight for quality assurance and would need to be closely managed.

Doctors in hospital medicine and primary care clearly articulated their need to know how to get further information about a disease or condition quickly. Interestingly, nurses did not feel the need for this, perhaps reflecting their role in more circumscribed conditions, where they could easily seek advice when faced with questions beyond their personal knowledge.

Whereas doctors were interested in the technical side of how to manage patients, nurses were keen to understand more about the family context of genetic disease. This would support them as providers of holistic care.

Doctors, nurses and dietitians all felt that they wanted to know more about gene testing, but interestingly, although mentioned, no groups thought that understanding more about predisposition testing was important at present.

Other areas were specific to the roles of the professionals: pharmacists wanted to learn about practical applications of genetics in pharmacology. Managers placed very highest priority on commissioning and particularly understanding about service models for genetics. They wanted to know what difference genetics could make to clinical management, as part of their need to make decisions about priorities for service developments.

Table 3, overleaf, details the priority given by each professional group to the areas of clinical and practical knowledge they would like to develop.

Table 3 Priority given to developing clinical and practical knowledge

Clinical and practical knowledge	Postgraduate medicine	Nursing	Dietitians	Pharmacists	Primary care	Managers
Conditions/clinical applications likely to see in practice (e.g. cancer)	**	***	**		***	
How to get further information and clinical support/understanding other roles	***		*	*	**	
Specialist genetics services		*			*	
Genetic screening programmes		*	*		*	***
Specialty based topics		*				
Cancer genetics	**					
Gene testing	*	*	**			
Family context of genetic disease		***				
Predisposition testing						
Genetic diagnosis and clinical implications						
Practical application of genetics in pharmacology				***		
How genetics informs clinical management						***
Service models for clinical genetics						**
Commissioning aspects						***
Genetic diseases that can be treated or modified	*					

(d) Supporting knowledge

Some professional groups identified basic knowledge that they felt they needed, that was particularly relevant if they were to become more proficient in genetics (see Table 4). Dietitians thought they would need to develop a basic understanding of risk, such concepts as relative risk, absolute risk, and even basic statistics if they were to be able to move into interpreting and communicating risk with patients. Managers and to a lesser extent pharmacists placed a high priority on developing their skills in evidence based medicine (EBM) and critical appraisal with particular relevance to genetics. Only managers mentioned the importance of understanding the national policy on genetics as a background to their decision making.

Table 4 Priority given to development of knowledge to support genetics proficiency

Supporting knowledge	Postgraduate medicine	Nursing	Dietitians	Pharmacists	Primary care	Managers
Basic understanding of risk			***			
IT skills						
Evidence based medicine and critical appraisal				*		***
Epidemiology			*			
National Policy on genetics						***

(e) Ethical, legal and social aspects

All groups mentioned the importance of ethical, legal and social aspects of genetics and, though this was not given the highest priority by many individuals, almost all individuals did think that these aspects should be included – which meant that, overall these aspects received moderate or high priority. This reflects clinicians' understanding that patients with genetic disease face a whole raft of social problems, including dealing with the familial aspects of disease and the implications of testing for employment and insurance, which would be significant factors in decision making and patient management. Also, that a thorough understanding of confidentiality as it relates to dealing with patients and their families, would be an important aspect of their clinical practice and of clinical governance.

Table 5 Priority given to ELSI aspects

Ethical, legal and social aspects	Postgraduate medicine	Nursing	Dietitians	Pharmacists	Primary care	Managers
ELSI aspects in general	**		***			**
Psychosocial aspects/multi-ethnic		***		*		*
Ethical/legal implications of testing		***		**	**	
Regulatory and policy framework				**		

Chapter 10 Policy points

Education in genetics must be based on learning needs arising from current realities of professional work, rather than on future possibilities

Professional groups vary in their emphasis but all clinicians include at least some molecular biology, clinical skills including family history taking and risk communication, how to get further information on genetics and ELSI aspects

II Genetics and genomics: preparing for genetics information in health and disease

II.1 Introduction

All stakeholder workshops were aware that the future of genetics was much wider than single-gene disorders (Box 9). Genetics and genomics would alter our understanding of health and the practice of medicine but it was not clear what the clinical applications would be and how professionals should prepare themselves for it. Most felt that it was premature to place great emphasis on this at present, particularly in CPD, where the motivation to learn from needs identified in clinical practice was a vital feature in effectiveness. At present there are no clinical applications for which professionals would have to change their clinical practice.

Nevertheless, there is broad support in the scientific and medical communities for the view that "molecular genetics will transform medicine in the twenty-first century". Furthermore, that the next five to ten years offer a window of opportunity during which health services can be prepared for an unprecedented development of new health technologies. Although there is debate about the degree of impact the Human Genome Project will have, (mostly concerned with scepticism over technical difficulties), there is considerable agreement that there will be developments in many areas of medicine with improved diagnosis, treatment and, most importantly, a shift towards greater ability to prevent common complex diseases. This paradigm change will depend on a shift in public and medical thinking towards one that places emphasis on human variation, different vulnerabilities to disease based on genes and environment, and increasing opportunities for prevention based on manipulation of the environment for the individual.

Most commentators do not believe that such a change will occur overnight. Nevertheless, the extent to which it becomes part of new practice will depend, not just on the eventual effectiveness of the new technologies, but also on the preparation of health professionals and their ability and willingness to understand and promote new possibilities and incorporate them into their practice.

In order to take this forward we present here an overview of the ways in which genetics and genomics will change medical practice, and some observations about the new clinical knowledge and skills that professionals will need to incorporate into their practice.

Box 9 Workshop participants recognised the wider implications of genetics for multi-factorial disease

Postgraduate workshop

Further into the future there will be a need to start preparing for "the uncertainty of how you assess multi-factorial disease and susceptibility loci." At present, we do not know when, and how useful, any of this will be, but professionals already have to advise patients on the basis of tests for multi-factorial disease available on the Internet.

"We need to have a competent understanding of what tests, therapies etc for genetic disease are available now, plus an openness of mind to what the world of genetics might offer in the future"

Nurse workshop

Nurses will increasingly be involved with patients at risk of adult onset disorders such as some inherited cancers where there is a strong genetic predisposition. In the future this will widen to include weaker predisposition to diseases such as Alzheimer's disease, diabetes and heart disease where there will be large numbers of individuals at risk who will request information from nurses in oncology, gerontology, general medicine and cardiology and, eventually most other specialties as well as primary care.

Nurses will be involved more in public health. "Public health nurses, health visitors and school nurses ... will need to be competent in genetics as one of the determinants of health."

Dietitians workshop

Nutrient gene interaction in disease is also an important research area with implications for the work of dietitians. It is a rapidly expanding research area and includes such diseases as cardiovascular disease, cancer, Alzheimer's Disease, infectious diseases and cancer. As well as genetic factors, nutrients also interact with each other and with smoking and drugs. Single gene polymorphisms (SNPs) may change the expression of a gene, possibly affecting the protein for which it codes in ways that might be important for disease.

It will be necessary to try and understand the scale and timescales for likely impact of genetics on clinical practice. Whereas the impact of single gene disorders such as PKU will be small, the impact of multi-factorial diseases such as diabetes and cancer will be much larger.

Dietitians should become competent in genetics for their future practice. The necessary education needs to be holistic, taking in biological and environmental aspects and would need to include statistics. Dietitians already take family history and have a general interest in this, but do not necessarily know what to do with it once taken with respect to advice using an understanding of genetics

Following these presentations of the progress and implications of research on genetics and diet, it was observed that dietitians currently may not know or understand enough to be able to interpret these findings and translate them into advice for the public. In order to "bridge the gap" between science and the public, dietitians would need to be able to understand such concepts as relative risk and odds ratio and associated statistics.

Pharmacists workshop

Pharmacists provide the key primary contact for most people with health questions. They must therefore be included as key health professionals who should be involved in realising the benefits of advances in genetics. Their role was, however, wider than simply providing advice and medicines on the high street.

“No matter how good and potent the medication, the ways in which the patient uses it and their understanding and belief about the disease has a fundamental influence on the process of restoration of health or amelioration of ill-health.”

As a guiding principle, therefore it must be recognised that pharmacists will play a key role in helping patients develop that sense of understanding of health and disease as it relates to genetics. This will mean that education moves further towards patient understanding and less basic science.

Primary care workshop

In topics grouped as ‘clinical knowledge’ genetics in relation to common diseases or situations was favoured. Thus the genetics of diseases commonly found in primary care, particularly ischaemic heart disease and cancers, were thought to be most useful.

Managers workshop

There is now a rare opportunity to see developments coming, and SHAs have a responsibility to have strategies that will anticipate future needs. Although there has been much hype predicting huge advances, for many managers there will be a degree of cynicism and resulting apathy in preparing for changes, particularly when there seem to be overwhelming other priorities.

11.2 Genetics and genomics in practice

With the completion of the sequence of the human genome a new set of challenges emerge as scientists move from enumeration of the sequence to exploration of the biological meaning of these vast quantities of data and the variations between individuals. The shift from genetics to genomics implies a shift from the study of single genes and their effects to the study of multiple genes, their functions and interactions. Medical and public health applications will change from a concept of disease in genetics, to one of information in genomics- the information that will result from one or multiple loci and strong interactions with environmental factors such as diet, drugs, infectious agents, chemicals, physical agents and behavioural factors. This genetics information can be used in diagnosis, treatment, prediction and prevention of all diseases and not just genetic disorders. It can be thought of more as a risk factor or biological marker than a disease state.

11.3 What will be the future applications?

In an article that reviews the implications of the move from genetics to genomics²³, Muin Khoury, of the Office of Genomics and Disease Prevention, CDC Atlanta, lists the following main applications arising from the Human Genome Project:

²³ Khoury MJ. Genetics and genomics in practice: The continuum from genetic disease to genetic information in health and disease. *Genet Med* 2003;5(4):261-268

Rapid progress in the diagnosis and management of single gene disorders, through genetic testing, and advances in medical interventions, including gene therapy.

Advances in gene mapping will lead to the discovery of additional disorders, most likely with incomplete penetrance (like haemochromatosis).

Identification and characterisation of numerous common genetic variants at multiple loci which increase or decrease the risks for various diseases singly and in combination with other genes and with various chemical, physical, infectious, pharmacological and social factors. This could be the basis for assessing disease susceptibility among health individuals, leading to personalised primary and secondary prevention strategies. Interventions included could be medical surveillance, lifestyle modifications, diet or drug therapy.

Genetic tests for individualised treatments including choice of drug and dosage - the science of pharmacogenetics.

Use of genetic information from somatic cells for diagnosis, classification, prediction and prognostication of many diseases, especially cancers, or for early diagnosis or screening.

Before any of these new technologies will be useful in clinical practice, much multi-disciplinary research will be needed bringing together basic sciences, clinical, epidemiological, behavioural, economic, health services, communications and outcomes research. This will provide answers to many of the current questions and in particular:

- How much is genetic information going to add to targeting interventions such as smoking cessation or hypercholesterolaemia?
- How much will genetic information add to what we already know from family history? Here professionals will need evidence based guidelines in using genetic information to profile disease risks based on analytical validity of testing; clinical validity and utility for improving outcomes and preventing disease; and assessment of ethical, legal and social issues for using such information.

The question for the research community will be the level of value-added to interventions based on genetic risk levels compared to prevention addressed at the general population

11.4 How should health professionals prepare themselves?

There is much that professionals need to do to start preparing themselves for the genetics era. A new way of thinking is required for both professionals and the public with whom they will work. This was described by Joseph McInerney in an article devoted to education for the public and professionals 'in a Genomic World'²⁴:

The contents of genetics education must change radically so that variation and individuality become the central message. The search for single nucleotide

²⁴ McInerney J. Education in a Genomic World. J Med Philos 2002;27:369-390

polymorphisms (SNPs) reminds us that genetics is the study of continuous biological variation - this is often obscured by a focus on single-gene disorders and deterministic treatments of molecular biology.

Perceptions of disease will have to change to overcome the assumption that all cases of a given disease are alike. The genome project and growing emphasis on proteins will take us nearer to underlying metabolic properties of a given disease and reveal, through the relationship between genetic differences and differences in protein product the individuality of disease in the form of vulnerabilities. This enhances prospects for prevention as a focus for health care.

In the same article McInerney lists some challenges for healthcare professionals as follows:

The explanation of causation is more difficult with common complex disorders than single-gene disorders. The doctor will not know the number of genes involved, nor their products nor how they interact, nor the unique ways in which experiences of environment precipitate disease.

With this uncertainty **determinations of risk and susceptibility are problematic** - risk from same gene product will differ from family to family and between individuals within families.

Where gene discovery makes susceptibility testing possible **counsellors will have to help decision making** - clear discussions about complex causation, the meaning of susceptibility and limited predictive value of positive or negative test results.

Professionals will be involved in the new area of **providing information about rationale for individualised treatments**.

11.5 What at this stage ought to be incorporated into education and how?

In the light of uncertainty about the detail of future applications, it is important, at present, to lay the foundations of knowledge and skills that will enable health professionals to eventually build genomics into their practice.

To begin the transformation from genetics to genomics, and the gradual shift towards prevention, basic education should be focussed more on genomics than on traditional medical genetics. Mendelian genetics should be taught as a subset of this where, in these diseases, the salience of one gene means that its products override the effects of products at other loci. The single gene usually has a profound effect on homeostasis since the gene is so deficient or dysfunctional that it badly damages the system of which it is a part. The relationship between gene, gene products and phenotype is much easier to discern and can be traced through families.

Health professionals will need to develop a more sophisticated understanding of susceptibility and disease that acknowledges human variation. They will need to incorporate these into their discussions of risk and options for prevention. To some extent it can be argued that professionals already do bring in concepts of inheritance through their use of family history in health promotion and disease prevention. As

the technology increases they will need to incorporate the use of genetic information into these discussions.

In their workshops, health professionals have rightly placed high priority on being able to assess genetic risk and communicate this to patients and help in informed decision making. Indeed, communication about risk and facilitating informed choice was the one area of clinical skills placed in the highest priority category by all professional groups. The breadth of this already covers both single gene disorders, and the risk that arises from family history indicative of familial susceptibilities (e.g. breast ovarian and colorectal cancer). Professionals indicated the importance of being able to use risk assessment tools and software that will assist them to provide advice on prevention options based on computer programmes containing information derived from large genetic databases.

Such skills will stand them in very good stead for the genomics era. As the technology advances and evidence-based genomic applications become available that can add further information to these deliberations, these will need to be supported into healthcare, as for all other technologies, with the necessary education and information.

Some practitioners will need to be involved in weighing evidence for themselves or for their organisations in particular areas of service development. Again, stakeholders recognised that they would need to develop their skills to do this. They identified that they would need to become better at evidence-based medicine - and able to use this in the area of genetics. The development of skills in epidemiology, critical appraisal, and concepts such as analytical validity of testing, clinical validity and utility for improving outcomes and preventing disease, and assessment of ethical, legal and social issues are all areas that have been identified by stakeholders. Whilst these are basic skills required throughout medical practice, these areas should receive particular consideration within teaching on genomics to ensure that issues pertinent to this field are incorporated.

Chapter 11 Policy points

The implications of the HGP would be that the emphasis for basic genetics education for all professionals should shift from an emphasis on medical genetics, and single-gene disorders to one that emphasises the molecular biology underlying human variation and disease.

The development of skills in risk assessment and communication should receive priority with a view to incorporating genetic information when supported by appropriate evidence.

Current computer assisted programmes for assessing risk and giving advice should be promoted so that professionals can gain experience of new ways of working in advance of further new technologies. These should be accompanied by appropriate education and support.

All professionals should receive education in evidence-based medicine. In each profession some individuals should become more expert in the issues arising from genomics and skills required for critical appraisal in genomics in their special area.

As new applications incorporating genomics become available, these will need to be supported by the appropriate education for professionals in practice based on an assessment of their need. The professional will need clear information to support use of the applications with an individual and to know how to access and use the information.

12 Resources for formal learning

12.1 Introduction

Just as people learn in a variety of ways, so the resources that they will use will need to cover a wide range, varying for different learning styles and the different aspects of knowledge and skills to be assimilated.

Professional stakeholder groups discussed and prioritised the sort of materials and programmes that they would like to have available. Summaries from stakeholder groups are presented in Table 6. The discussion below is augmented by findings from the review of medical undergraduate teaching undertaken for this project²⁵, the review of nurse teaching that formed part of the background work²⁶, findings from discussions with nursing colleagues in North America and an interim report on assessing learning needs for specialist registrars⁶ made available to the group.

Table 6 Priorities for educational resources

Educational materials	Postgraduate Medicine	Nurses	Dietitians	Pharmacists	Primary care	Managers
QA of resources					**	
Accessibility/database of resources with free access	**			***	**	
Library of case studies	**		*	*	**	
Scenarios for different themes in skills training (communication, critical appraisal)				**		
Videos of FH taking and consultations	*					
Worked examples of statistical risk			*			
Patient based scenarios		***				
Content appropriate to all professional groups					**	
A basic 'How to' resource pack				***		

²⁵ Jones, H. Owen H and Grant J. (an unpublished report) Survey of genetics teaching leads in medical schools. Open University Centre for Education in Medicine, May 2003

²⁶ Metcalfe, A (unpublished report). A survey of higher education institutions provision of education in genetics for post-registration and postgraduate nurses, midwives and health visitors in England, Wales, Scotland and Northern Ireland. 2002

Briefing notes and reading lists					***
Chapter in core text-book					***
Workbooks and guided portfolios		*			
Practical workshops					**
Patients as educators		**		*	
Simulated patient groups				*	
E-learning- supported by teaching faculty	**	***	***	**	***
Genetics issues in ethics and cultural awareness					**
Genetics in sexual health education					*
Bank of frequently asked questions				*	
Library of patient experiences	*			*	
CPD formal course				*	
Assessment bank	*				
Face to face learning of skills		**			
Genetics module in MSc				**	

12.2 What kind of materials are favoured?

(a) *Stimulating and illustrating with direct patient experience*

Accounts of genetic disease, and experiences of healthcare can have an astonishing effect on learning:

Nurses were often quite bored if genetics was presented just as basic science or as part of learning about disease processes. However, in the context of patients and their families whom nurses were looking after, they often became "transfixed" when they started to understand the impact this was having on families.

Nursing workshop report March 2003

Though these situations have to be carefully handled - sometimes learners can be quite upset by aspects of the accounts, and there is always a danger that the readiness of patients to disclose intimate personal feelings might be exploited - they are a very powerful learning tool. Resources such as video accounts, recordings or diaries can be used effectively as shown by websites such as DIPEX (experiences of health and illness) developed by the Department of Primary Health Care, University of Oxford [www.dipex.org].

As well as first-hand patient accounts, learning materials must include a substantial element of clinically based material such as case scenarios to work through, scenarios for training in skills such as communication, examples for critical appraisal, workbooks and guided portfolios, or resources for the incorporation of genetics into problem based learning. Didactic materials, in the form of lectures or

presentations would also have their place. Here the emphasis would be on the development of clear materials, which could be adapted by the lecturer and with supporting notes and advice on how sessions can be put together and how these sessions could be supported by local experts.

Materials must be presented in a variety of formats to suit different professionals' access to resources. For some professional groups the needs are for very basic paper-based material. For dietitians, a basic chapter on genetics in the core textbook would have the capacity to reach a large number of students. For pharmacists, basic information in the form of reading lists, briefing notes, and a simple resource pack would be a possible method.

All groups favoured some use of electronic learning. They recognised it as a useful backup and possibly the only means available to some professionals. However it should not replace face-to-face learning which would remain important to keep students motivated and to teach skills such as communications skills. E-learning would ideally be supported by a teaching faculty.

Those involved in education are also very aware that there is a lot of educational material on the Internet, but they often have no way of judging its quality. They would value materials that are supported by some sort of reviewing process or quality assurance. This is particularly important if we consider that material will have to be presented by teachers who are not necessarily expert in the subject.

(b) *The right materials for different groups*

The workshops revealed that education should be very context specific. It must be directed at the needs of the particular group and sub-group of professionals, such as cancer nurses, general practitioners, dermatologists, and pharmacists. Whilst professionals could see the usefulness of learning which crosses professional barriers, most felt lacking in confidence in this at present and expressed the need to learn as a single profession first, before adding in the multi-professional aspects. Where learning needs have been assessed, as, for example, in the work on learning needs of SpRs in non-genetics specialties, it becomes clear that there is an element of overlap in the more generic aspects, such as molecular genetics. However, the more clinically orientated areas and the specific examples used in each programme would need to relate to and be drawn from the specialist area (e.g. familial hypercholesterolaemia as an example of a single gene disorder in teaching of cardiology SpRs).

12.3 Developing materials

Formal educational programmes in genetics must be able to be delivered whatever the underlying structure of education. Work on the medical school programmes showed a variety of types of curriculum, including problem based, systems based, and topic based, or, more commonly, a mixture of all three. Genetics learning was covered, both as a separate subject, or integrated with other subjects to varying degrees. Methods of teaching included a mixture of lectures, small group teaching, self-directed learning, personal or group project work, problem based learning assignments, clinic attendance, discussion classes, laboratory or practical work, demonstrations and computer based learning.

The work of developing a full curriculum with supporting course material is very labour-intensive. At present, for the most part, teachers develop their own learning materials. Very little is shared, even within a department, and very few teachers access Internet material or CD-ROM.

Discussion in stakeholder groups underlined the perception that this is a relatively specialised field and that the process of developing the more sophisticated materials that fulfil modern expectations of learning and that fit in with the current methods of teaching will be quite an expert task. There are not many teachers in this area and few educational institutions that can cover the breadth of genetics. Thus, collective work to develop resources followed by dissemination, making resources available to other educational organisations and providing the necessary support to teachers was judged the best way forward for rapid and effective development.

The development of the curriculum to ensure quality and relevance will require what one of the stakeholder groups called 'expert groups'. Such work would be labour-intensive but would be necessary to ensure that, across the range, educational provision is developed that covers the breadth of the subject and does not simply reflect the expertise available in that establishment.

A framework for the expert groups to work on these programmes should be developed. Each would need, for example, to include a variety of stakeholders, including representatives from clinical practice, voluntary organisations, educationalists, experts in e-learning, geneticists, and experts in such areas as communication, ethical, legal or social aspects, depending on the areas to be covered and, most importantly, people who can represent the view of service users. Such a working group would then agree and confirm the key issues and topics to be covered, and develop a framework for each unit of learning and subsequent learning materials.

As such materials are developed, there is a lot of interest in being able to share by having them freely accessible and listed in a database, with sufficient supporting information to allow teachers to pick and choose. Our survey of genetics leads in medical schools showed a willingness to share and to participate in development. High importance was attributed to resources that were known to be regularly updated, formally validated by scientists, geneticists and other clinicians and where they were able to pick and choose topics, tailor to their own needs and style, and, in a climate where respondents had very limited budgets available, at very low cost or preferably free.

12.4 Organisational aspects for developing education programmes

Throughout the stakeholder workshops, we were made aware that there are certain structural prerequisites that would be advantageous or necessary to get educational programmes in genetics developed and implemented. These arose from the discussion of many of the barriers to development in the workshops. The organisational areas prioritised by stakeholder groups are given in Table 7.

Table 7 Priorities for organisational aspects for education

Educational resources required	Postgraduate Medicine	Nurses	Dietitians	Pharmacists	Primary care	Managers
Making information from Knowledge Parks accessible						***
National and local coordination for strategy and information	***	***		*		
Support of management framework						
Funding of all stages		***		***		
Free access to resources		***				
Regional coordinators/use of lead professionals	***	**	*		***	*
Learning facilitators	***					
Teacher time	***					
Support for tutors		**				*
Identifying sources of expertise				**		
Develop a cohort of teachers			**			
Better utilisation of genetics services for education					**	
Expert teams to develop educational programmes			***			
Protected time for learning	***					
Support networks for learning	**	*			*	**
Getting genetics into CPD			*	*		
Study update days/accredited	**					
Accredited training programmes	*		**		***	
Embed in existing training						***
Requirement in curriculum	***					
Multi-professional orientation			*	**		
Professional integration				*		
Developing multi-disciplinary education		**				**
Incentives for stakeholders				***		

(a) Funding

The education in genetics that takes place currently operates on barely adequate amounts of money. Developments in education have often happened because of personal enthusiasm rather than strategic and properly funded commissioned developments. This cannot be expected to be adequate if genetics education is to permeate throughout education including the doubters as well as the committed.

There needs to be funding of all stages: for groups of experts to agree and define competencies, develop curricula with resource materials, develop and support a whole cohort of teachers, provide incentives and protected time for professionals to learn, and provide finance for running courses of whatever description. Genetics teachers in most higher education establishments have little or no money for developing or gaining access to materials and rely on resources 'in-house' for their teaching.

(b) Coordination

Most groups felt that developing genetics education was a huge undertaking that would require many individuals and groups to be involved in defining competencies, developing educational materials and delivering education. The Strategy work had shown that there were many initiatives around the UK, for example the six genetics knowledge parks which all have roles in research, dissemination of information and education, and the many organisations, such as the Joint Committee for Medical Genetics, which has a role to promote genetics education, and the many providers of education.

For maximum effect, these various initiatives should be coordinated and a future programme based on close cooperation within and between these many professional groups.

(c) Local facilitation and lead professionals

All groups except pharmacists placed very strong emphasis on the use of professionals with special expertise in genetics and genetics education to stimulate and facilitate learning at a local level. Discussions in the workshops had noted the valuable role played by regional coordinators for the antenatal screening programmes and thought that this could be replicated more widely for genetics in general. Others noted that the development of GPs with a special interest or nurse counsellors providing outreach to local services provided a valuable source of expertise that could be used in learning. It is possible that something similar to this is already available to pharmacists in the form of their formal structure for continuing professional development.

(d) Developing a cohort of teachers

There is likely to be a deficiency in teaching faculty in the various higher education institutes across the country. A survey of nursing schools (see Box 10 below) showed that there were not many schools in which a full curriculum that covers all aspects of genetics could be delivered and that non-specialist teachers were being used to provide 'snippets' of genetics in the most relevant clinical areas. Schools did not express great confidence in their ability to teach the full genetics curriculum.

Box 10 Tutors providing nurse education in genetics in higher education institutions (HEIs) in the UK

A survey of HEIs providing education for post-registration and postgraduate nurses, midwives and health visitors found that only half (19/38) the institutions that supplied information on teaching of genetics to post registration and postgraduate nursing courses included genetics. Only the three specialist genetics courses had a full range of tutors with expertise in medical genetics, genetic counselling, ethics, law, laboratory aspects of genetics and epidemiology. In other courses qualifications of tutors varies widely with courses using PhD level scientists, genetic nurse specialists, nurses or midwives with a first degree in biology or a social science and laboratory based scientists. This implies that many nursing schools will struggle to teach genetics across the broad range unless their teaching faculty is developed first.

Alison Metcalfe, Hilary Burton March 2002²⁷

Those in postgraduate medicine particularly recognised that there would be a shortage of teacher time, a reflection of the very overstretched teaching commitments of the specialist genetics services at present. Nurses recognised that, as genetics needed to be taught in all nursing faculties, there would not be enough people with sufficient expertise at present and a group of teachers would need to be purposefully developed and then given strong support.

Pharmacists articulated this as the need to identify sources of expertise for teaching, and support to help genetics into parts of the course.

Teachers need support in “drawing students attention to the relevance of information derived from genetic technology and its interpretation”

Dietitians noted that there would be a need to develop a cohort of teachers and to put together a group of experts who will develop resources at a national level.

The need to develop a teaching faculty is a problem that should be tackled in some way. In North America there have been extensive programmes of ‘training the trainer’ whereby summer schools and distance learning courses have been set up for leaders from nursing faculties. One example of these is the *Genetics Program for Nursing Faculty (GPNF)* set up at the Cincinnati Children's Hospital Medical Centre. This has been funded by the Ethical, Legal and Social Implications Research Program of the National Human Genome Research Institute at the National Institutes of Health and the Division of Nursing, Health Resources and Services Administration. It includes Genetics Summer Institutes (GSI), Web-based Genetics Institutes, GSI participant follow-up, educational support and networking opportunities as well as two-day genetics update workshops every two years. Further details can be found on their website at www.gpnf.org.

²⁷ Metcalfe, A (unpublished report). A survey of higher education institutions provision of education in genetics for post-registration and postgraduate nurses, midwives and health visitors in England, Wales, Scotland and Northern Ireland. 2002

Such a programme required substantial funding, both for running the courses and for allowing faculty members to attend the residential course and participate in the web-based activities.

Identifying sources of expertise and developing a cohort of teachers was also recognised as important by pharmacists and dietitians.

(e) Getting genetics integrated into the curriculum

The process of curriculum change is complicated and can be quite slow. Curricula are full and finding room for additional content will be difficult.

Much has been said about the need to develop curricula at the undergraduate level to integrate genetics into education. However, in practice, the business of changing curriculum is a “slow, developmental, and iterative process”. There is a five-year accreditation cycle for courses.

Pharmacists workshop 28 March 2003

Some possible ways in which genetics could be integrated into the curriculum were discussed in workshops, including the following examples:

- Helping the faculty to become knowledgeable
- Locating genetics professionals who would act as a resource
- Identifying courses for potential placements - these might include targeting clinical component for discussion of genetics content/topic/issues - e.g. in obstetrics and paediatrics, taking a family history might be included in courses on history taking in general, or health assessment. Discussion of possible nursing roles in healthcare might be included in courses on obstetrics, paediatrics, psychiatric or community nursing.

(f) Structure of pharmacy training

A major problem for pharmacists will be the lack of a “properly structured and financed clinical placement model”. This means that it is difficult for students to develop skills in the application of genetics developments in practice, such as communication with patients about possible risks and benefits of genetic testing related to choice of medicines, until they actually emerge into practice.

(g) Support for learning

Learners and potential learners will need support to pursue educational needs in genetics. Groups recognised the importance of ensuring that genetics education could be recognised as formal CPD by making sure training programmes were accredited, and, in the case of pharmacists and dietitians, getting it into the formal programmes.

Protected time for learning about genetics, whether by including it in formal personal development plans, and, for a few individuals, special arrangements such as secondments or sabbaticals would also have a role to play, as would the setting up of local learning groups or support networks.

Nurses, managers, pharmacists and dietitians groups considered that the potential for them to involve practitioners in education about genetics would increase if educational groups were based on the multi-professional team, thus integrating their professional role within a care pathway.

Chapter 12 Policy Points

Learners and teachers need access to high quality learning materials including first-hand accounts and clinically based materials presented in a variety of formats

Educational material must be flexible enough to respond to individual needs.

Formal work to develop educational materials should take place on a national basis with materials freely accessible to all

The development of education in genetics will require substantial resources, development of teachers, support for learners, and local facilitation.

Opportunities for clinically based education in pharmacy education should be sought

13 Survey of medical undergraduate education

13.1 Introduction

The teaching of genetics to medical students was recognised as a priority by the British Society for Human Genetics. A group led by Professor Neva Haites from the University of Aberdeen undertook extensive work to develop an outline of essential core knowledge and skills for medical genetics. This was accepted by the Joint Committee on Medical Genetics in October 2002. The document lists 17 areas of core knowledge, and 17 learning objectives for clinical genetics²⁸.

The group recognised, however, that simply publishing such a list was not enough to produce change. As a next step, medical schools should be approached to find out how far these objectives were already in place, and to establish what support would be required across the UK to ensure that medical schools were able to develop their curricula to deliver agreed genetics education in the future.

A survey was undertaken by the Open University Centre for Education in Medicine in conjunction with the PHGU project. This aimed both to ascertain the current situation in medical education and to act as a paradigm for other areas in finding out the resources that would be required and the practical ways in which genetics education can be promoted and facilitated. A presentation on this work was given at the strategy building meeting and a report is available in full on the PHGU website at www.phgu.org.uk. Papers developed from this work will be submitted for publication in the medical literature in due course.

13.2 Summary of the work

Ten pilot interviews with medical school genetics leads were conducted to determine the content of a survey questionnaire. Leads in 24 medical schools received the survey and 21 (88%) participated.

Seven of the 17 essential core items, accepted by the Joint Committee on Medical Genetics were covered and assessed by the majority (50% plus) of medical schools:

- Chromosomal basis of inheritance
- Modes of inheritance
- Numerical chromosomal abnormalities
- Structural chromosomal abnormalities
- DNA as genetic material
- Types of DNA test

²⁸ Haites, N et al. Teaching Medical Genetics to Undergraduate Medical Students. 2002 Document agreed by British Society for Human Genetics and Joint Committee on Medical Genetics available at [\[www.bshg.org.uk/Official%20Docs/UNDERG~1.doc\]](http://www.bshg.org.uk/Official%20Docs/UNDERG~1.doc)

In clinical genetics seven of 16 essential items were covered and assessed by more than half the schools:

- Construct and interpret a family tree
- Recognise basic patterns of inheritance
- Risk of individuals suffering simple Mendelian disorders
- Clinical knowledge of several Mendelian Disorders
- Different forms of DNA testing
- Clinical knowledge of the genetic factors associated with cancer predisposition
- Genetic and environmental contribution to multi-factorial conditions.

A rich variety of curriculum models were employed ranging from problem-based learning, systems and topics based courses. Most were mixed. Genetics was quite commonly taught integrated with other subjects.

Lectures were most commonly used for the basic science teaching and there was also extensive use of small group teaching, self-directed learning and problem-based learning assignments. Clinical teaching included lectures, (81%), small group teaching (67%) and self directed learning (52%).

Materials for teaching were mostly resources that teachers developed themselves and were not shared at all (76%) although there was some development and sharing of materials 'in-house'. Only four schools accessed the Internet for material for basic science teaching and one (clinical) and two (basic science) used CD ROM based material.

When questioned about the future, three schools expected an increase in specific genetics teaching and seven intended more inclusion of genetics in other courses. The major barriers to change, however, were:

- Lack of time to develop resources
- An already overloaded curriculum
- Staffing levels
- Genetics not being seen as a priority by colleagues

In order to develop their teaching more than half the respondents said that they would like more staff, more curriculum time and a national resources centre. Over one third specifically said they would like links with genetics facilitators and access to teaching materials.

They would specifically like an updated list of resources for teaching on the Web, centrally developed curriculum resources and particularly problem-based material, case histories and histories that illustrate ethical issues.

These resources would have to be known to be regularly updated, formally validated by scientists, geneticists and other clinicians. There would have to be flexibility to pick and choose topics and adapt them to a particular course, relevant to the core curriculum, and most importantly free because schools have no or limited budgets.

In terms of how resources might be developed, the survey showed that medical school genetics leads would be willing to be involved in national development of genetics education either as active contributors (11), reviewing or piloting materials (14), contributing resources already available (11) or as users, or observers.

On the basis of this review, some recommendations were made about national developments.

- Any national development must be flexible enough to respond to local conditions and requirements
- Any national initiative must not put further demands on medical schools or teachers but should support their current arrangements
- A national resource centre, genetics facilitators and flexible educational resources should be established. This should include a regularly updated list of web-based resources, problem based resources, case histories and ethical challenges
- All nationally provided or recommended materials should be quality assured
- Any national development should ensure that medical genetics leads are fully involved in roles that suit their availability, interest and expertise

13.3 Discussion

These findings substantiate, through their more rigorous approach, much of the discussions from the workshops about the teaching of genetics, difficulties and barriers to development and the resources and support that would be valued. Most importantly they support the view for a way forward in which teachers had more national support and were able to share experience and access the more complex teaching materials from a central resource and adapt them for local use. The willingness of teachers and experts to cooperate on initiatives to achieve this was encouraging, but it is clear that such work would need to be negotiated as part of teaching time and not simply an extra burden. Further, any joint initiative would succeed only on the basis of the known quality of support to teaching and the extent to which resources were accessible.

Chapter 13 Policy Points

Development of nationally agreed core competencies for all professionals should be the starting point for influencing the curriculum and deciding on necessary learning materials

Further work to increase depth and breadth of genetics within the medical undergraduate curriculum should be undertaken

There is much support for developing and sharing complex materials on a national basis

14 Learning in a professional environment

14.1 Introduction

Whilst education of undergraduates and those in specialist training is important for preparing the future generations of health professionals, the stakeholder groups recognised that there was a major task to be undertaken to develop genetics competence in the 'prevalent pool' of practitioners, those undertaking today's service. The groups all considered continuing professional development to be of prime importance, placing emphasis on **raising awareness** about individual and organisational learning needs, **formal and informal education**, and the support needed to enhance **learning from clinical practice**.

14.2 Education in genetics within managed CPD

A formal programme to enhance competence in genetics should be embedded firmly within the framework for CPD as set out in *A First Class Service*²⁹ and in the guidance about CPD in general practice. This framework was developed on the basis of evidence of effectiveness of CPD and on the professional needs and approaches to continuing education. It can be used for all members of the care team. Most importantly, the framework places CPD within a management process that supports it and makes sure it is relevant, and in doing so enhances its effectiveness. In this way, CPD is seen to enhance quality and is one of the main areas through which an organisation ensures clinical governance.

It follows, therefore, that until genetics aspects of mainstream services are recognised as key aspects of high quality services, it will be hard for individual professionals and the organisations for which they work to argue strongly for their inclusion in formal professional development. Further, until these sorts of mechanisms are built into organisation plans, we will have very little way of knowing what professional development has been undertaken, what particular aspects of genetics are priorities and what CPD for genetics is effective.

The document *The Good CPD Guide*³⁰ goes on to detail the formal processes that professionals and their employing organisations go through. This follows a cyclical process first to identify need for CPD, prepare personal development plans, undertake CPD and then reinforce and disseminate it to show its effectiveness and finally monitor the whole personal development plan.

²⁹ A first class service. Quality in the new NHS. Department of Health. 1998.

³⁰ Grant J, Chambers E. The good CPD guide. A practical guide to managed CPD. The Joint Centre for Education in Medicine, 1999

14.3 Processes in professional development

Needs assessment is the first critical step in CPD and is the process through which a professional or organisation recognises a learning need. This is firmly linked to the findings of the stakeholder workshops that raising awareness of genetics is a vital first stage. Unless professionals recognise that they have educational needs, they will not be motivated to learn.

The Good CPD Guide lists many ways in which professionals become aware of their own learning needs that are useful both in the formal CPD programme and informally. Some occur simply as part of the normal routine of clinical work, and others require a more conscious effort, but all could be directed towards revealing unmet needs in genetics.

- The clinician's own experience in direct patient care
- Interactions within the clinical team and department
- Non-clinical activities
- Formal approaches to audit and risk assessment
- Specific activities directed at needs assessment
- Peer review

14.4 Learning methods

The methods through which professionals learn are diverse and the document acknowledges that professionals live in a learning rich environment and there is no one best or 'right' way of learning. Much of their learning is integrated with their practice and often arises from it; a style of learning known as 'situated learning'.

The CPD guide describes how much of the learning of professionals follows the professional apprenticeship style, applicable to juniors and also used by seniors, who might include, for example, discussions of patients with colleagues. It includes such elements as experience of seeing patients, building up personal knowledge and experience, discussing patients, listening to experts' explanations, 'bite-size' learning and learning from teamwork interactions.

Many of these methods rely on access to authoritative information or to those with more experience. In the context of genetics, stakeholders stressed the importance of validated information or guidelines on the Internet, access to genetics specialists, either in a Centre, or as outreach from the Centre, and access to fellow professionals or team members with some more special expertise in genetics, such as perhaps a specialist health visitor for children with special needs, a coordinator for antenatal genetics screening programmes or practitioners (GPs or nurses) with a special interest in genetics.

The nature of specific CPD interventions in changing or developing practice is less important than the fact that they should be multiple and targeted on professional needs.

The many different formal and informal learning methods that professionals use are detailed in the *Good CPD Guide* and fall into the following categories:

Academic activities - including research, reading, writing and revising service and research protocols.

Meetings - department meetings, courses, lectures, workshops.

Learning from colleagues - for example, other team members, formally or informally, consulting other professionals.

Learning from practice - the basis of continuing expertise necessary to think about practice can use formal and informal ways - e.g. diaries, use of evidence based reviews, learning from the ever-increasing range of patients and their outcomes learning from experience and applying what they have learnt to future practice.

Technology-based - audio-visual, communication and information technologies, computer support systems (e.g. information base and guidelines for active decisions), distance learning.

Management and quality processes - use of accreditation, audit, inspection visits.

Specially arranged opportunities - such as attachments and secondments, and sabbaticals.

All of these are areas that would need to be built upon as CPD developed to include genetics.

Chapter 14 Policy Points

Inclusion of genetics in CPD will be increased when genetics is regarded as an important aspect of high quality services

An educational programme will need to actively pursue ways of making professionals aware of their own learning needs

A CPD programme for genetics should work through a range of learning methods to suit different learning styles and access

15 The development of the Internet to provide information and guides to local services

15.1 How well placed is the Internet in the UK to provide clinical support?

There is a lot of information about genetics on the Internet. Sites were detailed in the background document and cover such areas as basic genetics and genomics, genetics and disease, genetics epidemiology, public health genetics, genetics policy in the UK and ethical, legal and social issues. However, we were aware that much of this was US-based, and for most it would not necessarily be obvious to health professionals how to find it or whether it was authoritative.

We also learned from the workshops that professionals need very rapid access to information and guidance on services in support of their clinical care. If specific queries could lead them on to access more formal education in genetics, this would also be an advantage.

We were aware that a number of websites had been set up in the UK by genetics groups to provide such a service for health professionals. Foremost among these, and probably the obvious first port of call for health professionals needing local information for their patients were the regional medical genetics centres. The BSHG also has a website which represents groups of professionals with an interest in genetics: clinical geneticists, cytogeneticists, molecular geneticists and genetic counsellors. There were also rudimentary websites that had collected information on genetics such as those within the RCGP website and the National electronic Library for Health.

We wondered what lessons could be learned from an evaluation of these websites about the sort of information that was useful to professionals, the ways in which it could be made accessible on a national basis and, ultimately how it could be used to provide an entry point to wider educational material.

As part of the strategic work a review of these websites was undertaken by Mike Greenwood, an independent media analyst. The full report of the review is given on the PHGU website at www.phgu.org.uk.

15.2 Main findings

Needs expressed by professional users included:

- Educational information about common conditions
- Information about rarer conditions which is easily accessible and known to be reliable and up-to date and can be used in an immediate clinical context, such as that on haemoglobinopathies which is available on the APoGI website www.chime.ucl.ac.uk/APoGI/menu.htm

- Practical resources to support clinical management, including, for example, tools to construct and record family pedigrees, guidelines for referrals, practical information about what the patient can expect from the whole referral process
- Information sheets about clinical conditions to use with patients
- Practical support to help understand wider legal and ethical issues which may be confronted in caring for given patients

To what extent are current websites able to fulfil these aims? Provision varies, with only about 50% of services having any significant web presence, but these are of very varying depth and quality. The websites can be hard to find, the available list from BSHG or GIG tend to duplicate mistakes or omissions. Their position reflects complicated organisational structures, for example as parts of university or hospital websites or sometimes freestanding.

There is a range of objectives and target audiences. The focus may be primarily patient, professional or academic. There are also differing levels of ambition, and resources. Some simple sites offer useful information on the physical resources on offer – clinic times and locations, contacts, help with referrals process. This was thought to be very useful. Other sites create deeper, structured content, such as much more theoretical consideration of genetics, but seem to be doing so independently, with duplicated effort.

The information on the website rarely provides an integrated service, ‘seamless’ for the user. Relevant content is sometimes fragmented and hard to find. Not all sites have a search facility to find relevant content that might be there. Thus, for example, information on hereditary cancers in the cancer part of a hospital website may not be linked with the genetics department who would provide a service for genetic testing. The quality of links both within such sites and with other supporting information available on the Worldwide Web varies considerably.

Some content is directed at GPs, but there is little evidence of material aimed at other groups of healthcare professionals such as nurses or midwives.

Some useful resources are evident. Examples include support for good clinical practice, for example, how to take a family history, understanding and explaining risk, Frequently Asked Questions (FAQs) for use with patients and their families, knowledge of the range of tests available and their processes, up-to-date information on therapies available, peer exchange, downloadable, reusable resources such as care cards and online guidelines and referrals forms, and fact sheets on medical conditions.

There is very little formal educational use of the web –either to provide information about courses or to offer web-based modules, for example as part of continuing professional development. There would be great scope for this by providing access points to the many educational resources available on both sides of the Atlantic, as summarised in the background document. There would be more scope for covering the social, cultural, ethical, and legal implications of genetics.

Finally, maintenance is an issue with sites often being difficult to keep up to date because of lack of resources, or complex bureaucracies within the parent organisation.

Thus, whilst there is a lot of good work being done with websites, probably more could be achieved more efficiently by some coordination of effort. This might be difficult to achieve. However, some central facilitating and development of content and perhaps the development of a template which departments could use as they saw fit would be helpful. The possible role of the NeLH in providing some centrally agreed content and access to current educational materials is not yet clear.

15.3 The way forward for Internet resources

In the Hinxton workshop a session was devoted to considering the way forward for developing Internet resources across the UK. The need to provide information, clinical support and access to education was agreed, built on the principle of a patient centred service that would provide information to professionals to enable them to provide effective care for patients. It was recognised that current provision was piecemeal. Whilst there were pockets of good practice, provision was far from universal, with some departments having no websites. Most were the product of individual enthusiasm rather than reflecting organisational commitment. This was reflected in the quality of websites. When time and money were scarce websites were not a high priority and often became out of date and the development of new material was not sustained. There has also been much duplication of effort in the development of generic material.

Through discussion with website providers and users at the workshop we tried to find a way of building on and encouraging the work of enthusiasts whilst getting others to basic levels and using the whole system to harness the many excellent resources available on the worldwide web. It was accepted that improvements in local clinical support could be achieved by working together, sharing the development of generic materials and supporting regional departments to develop websites that related to their own local services and particular specialist interests. However, those with more advanced websites now would want to retain their independence and ability to develop further according to local needs. The provision of educational materials for local undergraduate medical courses and guidelines and information for particular specialist areas were good examples.

Three models for the overall provision were considered:

- Centralised: a single portal, organised and accessed centrally
- Decentralised: very local and left to local enthusiasm
- Third way –core resource linked to local resources

The third model was the favoured model because it combined local flexibility and resourcefulness with national support for local arrangements.

It was recommended that the organisation of such resources should include both 'top down' and 'bottom up' elements. It would be developed as a self-organising,

democratic community. There would be a small central organisation providing support and a range of linked local sites with entry to the whole network being via any of the local sites. The central information portal would be accessible to both professionals and patients and it would provide links to educational resources worldwide, regional departments, disease-specific areas and specialised areas (such as the Scottish Muscle Network).

The functions of the central resources would be:

- To provide IT technical support
- To commission local and specialist content sites
- Validation and accreditation
- Coordination
- Host for some material
- Organisation and administration

A small central team with local involvement/representation would:

- Write templates for local sites
- Provide pump priming finance
- Establish links between information and education and between sites
- Commission providers to fill gaps in available information content.

Chapter 15 Policy point

The Internet will be a valuable resource for clinical support and education and the Regional Genetics Departments, NeLH and other specialist genetics websites should be developed to provide clinical support, information on genetics diseases, and information about services for all health professionals as well as access to genetics education on the worldwide web.

16 The use of service developments to promote genetics education

If we are to get genetics education built into NHS services one of the most potent times to do it will be when services are making developments.

16.1 Introduction

Clinical care is based on teamwork. The most complex clinical pathways will involve many individuals, from the receptionist who makes the initial appointment, through to the many specialists or practitioners who might eventually be involved in continuing care. It may involve many sectors, from primary care to tertiary, and many different clinical areas, often including pathology and radiology. Such complex systems rely on professionals being competent in their own roles and also having an understanding of the roles of others in the pathway of care. Experience, such as that in the cancer collaborative programme [www.modern.nhs.uk/cancer], has shown that great improvements in outcome and the efficiency of services can be achieved by taking apart and examining in detail the processes of care. Learning can be transferred from one centre to another and in this way overall service improvements can be achieved.

As services develop, staff must learn to do things differently. It is a responsibility of those commissioning and providing new services, or service elements, to ensure that these new competencies are developed within the implementation programme. The many areas where genetics is becoming a factor in such new services offers a formal opportunity to consider the educational needs of practitioners and the ways in which they could be met.

16.2 Opportunities for genetics education in service developments

We considered how we could actively use current and planned service developments to incorporate an element of genetics education by examining required roles in detail and considering what competencies, and hence what learning might be required of the workforce. Some current developments where opportunities would arise were discussed in the Hinxton Workshop and are given in Box 11, overleaf:

Box 11 Service developments where genetics education may be an important factor in implementation

- *New national screening antenatal and neonatal genetic screening programmes for haemoglobinopathies, Down syndrome. Education is currently being commissioned in support of these programmes*
- *National Screening Programme pilot studies such as those on colorectal cancer*
- *The development of cancer networks. There are many opportunities to consider what competencies are required to ensure the programmes deal competently with hereditary and familial cancers*
- *The development and subsequent implementation of guidelines for these with a family history of breast cancer – currently under consideration by NICE*
- *The development and implementation of decision support software for people with a family history of breast and other cancers*
- *Development of coronary heart disease networks*
- *Pilot studies for the implementation of cascade screening for family members of people with familial hypercholesterolaemia*
- *Developments in pharmacogenetics*
- *The availability of over the counter genetic tests*
- *New genetic tests. The genetic testing network has developed a procedure for the evaluation of potential new genetic tests. The process of implementation could include a formal consideration and evaluation of the education requirement for those involved in offering the test and others in the care pathway.*
- *Enzyme replacement therapy*

Whereas some of this development of education through service development would take place on a national basis, further opportunities could be sought to work locally with some services developing their Local Development Plans. These include planned and unplanned developments and are required to have parallel workforce development plans. The way in which these might consider and include genetics within these plans could be pursued.

Chapter 16 Policy Point

Opportunities should be systematically sought to tie genetics education to both current and future service development where genetics is a significant component of care pathways

17 Developing ownership for the educational programme

17.1 What do we mean by ownership?

The theme of ownership is one that recurs throughout the strategy development process and was one of the major topics in the final workshop.

Ownership of the strategy, programme, educational curricula and educational materials and information that will be developed are important elements for success. Whilst it may not be too difficult to get organisations to sign up to the work, real ownership will imply that they feel that this is their programme, that they were involved in its development and that they want to use it.

Work with stakeholder groups and the review of medical undergraduate education has clearly told us that there is a need for and interest in educational resources produced nationally for local use, that there is a need for access to these and other resources, and that there would be support for a national organisation to coordinate and enable this. We also know that there is a willingness of educators and specialists to be involved in the development of materials, limitations being mainly pressure of work rather than any difficulties in attitude or principle.

What is clear from our discussions is that ownership is not 'all or nothing' and cannot be obtained all at once, or by the sheer logic of the argument. It is something that will be gained gradually, at many different levels by the processes of inclusion, involvement, communication and the production and dissemination of very good work.

17.2 Methods for gaining ownership

The sheer complexity and decentralisation of health services mean that there is no single route to ownership. We identified three main levels. At the top the statutory bodies such as the General Medical Council, and Nursing and Midwifery Council and the many professional royal colleges all have a role in setting the general outlines for the syllabus to be followed. It is important that these bodies are persuaded to include some elements of genetics in these syllabi so that it will be included in accreditation and inspection processes.

These will not be expected to include a great level of detail on the depth and range of genetics to be included. The next level is therefore that of the professionals who are involved in the delivery of education. Until recently this has been left to individual education programmes, with much variation in content from place to place. However, there are advantages to be gained in working together on such things as core competencies and learning objectives in terms of the strength or authority it gives to arguing for genetics teaching resources within a programme. We heard an example of how learning objectives were agreed for medical undergraduates during a two-year process led by Professor Neva Haites. This process involved hugely committed leadership, the involvement of many members of

the British Society for Human Genetics, national meetings, much e-mail correspondence, reviewing of draft documents and discussion for individuals within their own medical school before a final document was agreed. Nevertheless, the strength of this consensus has enabled developments in the medical schools because it has shown a clear view of the expected standard. A similar process is now underway, led by Dr Maggie Kirk, to develop core competencies for nurses. The key messages here to develop ownership are *leadership, inclusion and involvement*.

Whilst medical schools and deaneries are important for promoting genetics in medical education, the Workforce Development Confederations play a lead role in commissioning education for nurses and all other healthcare workers. Their support will be crucial in managing programmes into place at the local level and it will therefore be important that, as a group, they gain ownership of the programme. The Chief Executives work well together and have mechanisms to share developmental work, which the programme should exploit, to gain involvement. Thus we might seek involvement of a representative of the WDCs to develop and implement a particular programme. As well as developing an overview of programme content and the development process, WDCs could use their particular expertise to advice on how to commission the programme through contracting and other mechanisms.

Ownership must be achieved for the individual elements of the teaching programmes. The key to this is transparency and inclusiveness of the process of programme development. Teachers might be persuaded to use the material if they can see that it has been accredited and quality assured in some way. This helps them to be more accountable when they are visited and inspected. However, ownership will be improved when people can adapt national material for local needs – and pilot material should be developed that has both educational content and pointers to the ways in which local experts, both patient and professional, might be used to illustrate some points or lead some of the discussions. There will certainly be some element of training the trainer or cascade training to enable this to happen, and this is another good way of promoting ownership of nationally developed programme at local level. An example of the ways in which pilot material might be developed and delivered is to try and use PCT protected learning time for the primary care team to focus on genetics. This sort of educational outreach is being piloted in Portsmouth and outline programmes developed. However, it is not clear whether there would be enough experts to carry out the teaching. It would be necessary for the professional groups to have product champions at local level and many stakeholder groups mentioned the importance of local clinicians with a special interest in genetics, or local facilitators for learning.

Finally, ownership must be achieved for the individual professional. This comes back to raising awareness and then being able to deliver education that seems relevant to their particular clinical practice. It will require skilled teaching and mentoring and the intelligent use of scarce resources such as videos of patient interactions.

Chapter 17 Policy Points

Ownership of the Programme should be sought throughout the service, including:

- **Statutory bodies and professional associations**
- **Higher education institutions and other education providers and commissioners**
- **At a local level including those in everyday practice**

18 The role of the pharmaceutical sector

18.1 A survey of genetics education in the pharmaceutical industry

A survey was undertaken to develop an overview of pharmaceutical industry's education infrastructure and to establish current and future education and training plans in field of genetics for health professionals and the public. This involved an initial web-based search, followed up by in-depth interviews with 8 selected companies.

18.2 Main findings

The initial Internet search of 20 companies showed that most education is orientated towards patients, is disease-based and reflects products. Not all companies have web-based professional education and genetics education and information is almost non-existent in the UK.

The usual pattern is that education does not have dedicated company wide resources but is product based and delivered through the brand team. Usually the education relates to diseases and product knowledge, but sometimes skills based training not linked to brands but linked to the NHS modernisation agenda such as business and time management training is offered.

All companies thought that education and training in genetics for health professionals was essential but activity so far in the UK is limited. Companies had funded some education for health professionals in relation to genetics, but this was mainly limited to genetics exhibits and presentations. However, GSK Genetic Research Group had dedicated resources for running genetics education programmes, both internally and externally.

The key competencies that companies thought professionals would need to develop were:

- Communication
- Counselling
- Medical and patient information resources
- How to read a family history
- Genetics background and terminology
- Current genetic science
- Informed consent
- Pharmacogenetics and its applications
- How genetics can be used in practice

Most importantly, the need for education for health professionals was endorsed by all participants, and all were willing to participate further in discussions to develop a co-ordinated approach.

Chapter 18 Policy Point

Partnership from the pharmaceutical sector should be sought in the development of an educational programme

19 Experience from North America.

19.1 Project visit to NCHPEG organisation

Four members of the Group attended the National Coalition of Health Professionals Education in Genetics (NCHPEG) Conference in Washington in January 2003 and had talks with the director of the Coalition and other delegates, concentrating on what factors had been critical in the development and success of the organisation and pursuing particular areas of interest such as primary care, and looking at methods for delivering genetics education or clinical support.

19.2 The development of the Coalition

The Director of NCHPEG, Dr Joseph McInerney gave an overview of NCHPEG from its beginnings in 1997.

One of the most critical factors in the establishment of NCHPEG had been a high level of ownership across professional groups. The coalition was originally promoted by Frances Collins, the Director of the National Human Genome Research Institute. As a physician he was able to see the implications of the Human Genome Project for health and healthcare and he was able to use his influence to recruit health professionals from the tops of all the professional associations.

It was also important to involve the specialist genetics community and to have opinion leaders in each profession. The main problem had been how to get other professionals involved and interested in genetics. The main message to achieve this was the ubiquity of genetics in medicine and the huge momentum for genetics being built up through research.

A great danger in all this was the risk of overselling genetics. They had learnt that **professionals are mainly interested in what they need to do differently in their own practice.**

The main areas of work for the Coalition so far were:

- Development and evaluation of core competencies
- Annual conference and speciality on-line conference
- Genetics Resources on the Web (GROW)
- Development of work on family history
- Development of educational programmes (psychiatry, common diseases, pharmacogenomics)

19.3 Primary care

Jon Emery provided the main link with primary care genetics and presented a report that assessed the possible usefulness of closer working in this area.

NCHPEG has developed a list of core competencies in genetics, listing 17 knowledge points, 17 skills and 10 attitudes. These would appear to be an 'idealistic view' of what we should be aiming for in UK, although they are listed as core competencies in North America. They are thus very ambitious and would be a good starting point from which to develop curricula in this country.

The Centre for Disease Control (CDC) has identified family history as a key area to develop both family history-based skills and tools for managing chronic disease in primary care. They are funding a number of programs over the next 3 years that will be assessing different ways of supporting the management of people with family histories of common chronic disease. Progress so far has included family history questionnaires and guidelines, all paper-based.

Over the last three years the *Genetics in Primary Care* programme³¹ that was funded by various national organisations has developed a series of curricula materials to be used by primary care faculties. This included the training of residents, and support for primary care practitioners across various faculties throughout the US. The Programme aims to provide useful case based materials to support teaching across a wide range of areas. Examples include breast, ovarian, colorectal cancer, cardiovascular disease, congenital hearing loss and dementia. The important ethical, legal and social issues are incorporated into each. All the materials are available on-line, but it is anticipated that teachers will incorporate it into their existing teaching programmes and certain teachers in each faculty will be supported to develop it. There is currently some evaluation of this undertaken by the University of Wisconsin, but none has yet been published.

If we are to avoid 'reinventing the wheel' for the UK these materials could provide a very useful starting point for UK specific materials. The importance of building on what was already available, rather than totally developing our own was underlined by the expensive nature of these developments in the United States. For example, a programme to develop a CD-ROM on psychiatric genetics, recently undertaken by NCHPEG had cost \$680,000 and their current programme for the genetics of common diseases had received a grant of \$792,000. These programmes are therefore not cheap to undertake.

The teaching materials would need some UK adaptation and those responsible for them in the US have indicated that they would be very happy for them to be adapted for the UK and suitable for an NHS setting. This could therefore be very useful for the future development of a UK programme.

³¹ Burke W, Acheson L et al. *Genetics in Primary Care: A USA Faculty Development Initiative* Community Genet 2002;5:138-146

This adaptation would need to take into account the differences in family practice in the US. For example there is direct access to many genetic tests for primary care physicians and one of the main incentives for education has been that the US is trying to stem the potential misuse or mis-ordering of genetic tests directly by primary care doctors – a group which includes internal medicine and paediatrics as well as family practice. Thus, they are less focussed on the gatekeeper role than in the UK.

Furthermore, specialist genetics services are actually very patchy in the US and a number of the Health Maintenance Organisations and private insurance companies do not subscribe to genetics services. Thus primary care and other non-genetic specialists are the only practitioners available for genetics advice and services and so they may require greater knowledge and skills than practitioners in the UK.

19.4 Developments in genetics education

The Conference provided many examples of genetics education programmes or ways in which health professionals could be supported to give better genetics advice. Some of these are listed in Table 8.

Table 8 Developments in education in the US – some examples

Organisation	Development
National Organisation of Nurse Practitioner Faculties	Work undertaken on strategies for integration of genetics into nurse practitioner education
Maimonides Medical Center New York	Use of simulated patients to teach
Human Genome Education Model project (HuGEM)	Approach to teaching 7 professional groups targeting administrators, leader educators and practitioners using orientation sessions, weeklong core courses and training workshops.
March of Dimes	FirstPAGE A resource to provide genetics education and guidance to prenatal care providers
Genetics in Primary Care Workgroup	Products developed on family history taking, 'red flags', cultural competency and evidence-based medicine
NIH, NHGRI and Office of Rare Disease	Genetic and Rare Diseases Information Centre A centre staffed with information specialists to answer questions for professionals and patients
Cincinnati Children's Hospital	A web-based genetics institute for nursing faculty
NHGRI and NIH	Web-based self-educational modules with a monthly lecture given by nationally recognised genetics expert and an e-classroom for questions, testing understanding and discussion
Foundation for Blood Research, Maine	A CPD programme for public health nurses including a Genetics Resource Guide and annual regional CPD conference offering CPD credits

This provides only a sample of work being presented in the US, but is an indicator of the large amount of detailed work that has happened in the last few years to develop genetics education for different groups of professionals and using different methods. Though any approach would need to be adapted for the UK, it is recommended that close relationships with US and NCHPEG should be built up so that we can learn from this innovative work, build on this experience and, in the longer term, work with colleagues in US to develop further applications.

Some prime examples for early work might be:

- Education in primary care
- Educational work to develop nursing faculties
- Tools to support professionals providing antenatal services to identify and act upon significant family history or genetic information
- The use of electronic 'introduction to genetics packages' for health professionals
- Information services on genetic diseases
- Exploring educational needs and approaches for other groups such as dietitians, physiotherapists, occupational therapists and social workers

Chapter 19 Policy Point

The UK should seek alliance with NCHPEG in developing its educational programme in order to build on the work already achieved in the US and in the longer term to work collaboratively on the development of further educational resources.

Section Three

Conclusion

20 Conclusion

In 2000 the Public Health Genetics Unit collaborated with the Nuffield Trust on a major project to examine the potential impact on the UK's health services of the "remarkable developments that are taking place in the field of human genetics"³². Genetics education for professionals and the public was identified as one of the key issues that would influence outcome and, in particular, our ability to capitalise on advances in genetics to improve human health in this country.

The following year, the Unit was asked to take the lead in establishing current educational needs and provision, and subsequently to work with stakeholders from professional groups and the public to develop an outline strategic programme that would begin to prepare the health workforce to meet new challenges in genetics.

This report is the culmination of that process. In such a wide area we could not hope to have compiled a complete record of all the many initiatives nor to have spoken to all those with interest or expertise. However, we have drawn together a wide-ranging group of well-informed contributors. More than twenty patients and their families, experts in the experience of genetic disease, met us to give their views and a further thirty gave written evidence. Eighty professional experts with various viewpoints attended workshops. We were impressed by the ability and commitment of them all. Particularly exciting was the ability of those who had previously given little thought to genetics to embrace new concepts, to understand the relevance for health services and to consider the implications and possibilities from their own viewpoint. These people - teachers, commissioners, leaders of professional associations - also demonstrated a strong commitment to taking genetics forward in the UK.

We have undertaken detailed reviews in a number of key areas - medical undergraduate education, postgraduate nurse education, the roles of the Internet in the UK and the potential for collaboration in education amongst pharmaceutical companies. We have established contact with colleagues in the US and sought advice from those with practical experience in developing national genetics education programmes.

All our evidence has been sifted, reviewed and discussed at length and, consequently, we have made proposals for a programme which could be implemented in the UK. We believe this programme best combines our understanding of what works with the human resources, energies and enthusiasms that we have available.

We are confident in having established the optimum way forward and perceive no advantage to further wide scale research. There are no 'right answers' to get closer to nor more exact verdicts to be reached. Importantly, we recognise that the

³² Zimmern R and Cook C. Nuffield Trust Genetics Scenario Project. Genetics and Health – Policy issues for genetic science and their implications for health and health services. London: The Stationary Office, 2000.

progress of the programme will not be set in stone, indeed, its ongoing and dynamic nature will be core to its success. Many details, such as the content of curricula, are still to be worked out, and there must be an integral process of review and evaluation that will continually guide the advancement of our strategies. In the ever-advancing field of genetics, such safeguards should give us the confidence to begin a programme with the knowledge that we can be responsive to change. This flexibility is crucial as new applications come about, as professionals undertake new roles, and as we gain experience of the impact of our education programme.

We believe that the time has now come to implement our recommendations and, for this, the announcements in the Genetics White Paper provide a very welcome catalyst. We should move forward in a partnership that acknowledges the interests of the many organisations in this process, from the researchers keen to see the translation of their work into health benefits to those involved in the delivery of health services, and from public health specialists to the private sector.

In 2001 the Secretary of State announced a new ambition for Britain: to put us at the leading edge of advances in genetic technologies and to develop in the UK a modern genetics health service unrivalled in the world. A competent workforce will be fundamental to achieving that vision.

Section Four

Appendices

Appendix I: List of participants

Mr Gerald Alexander	Community Pharmacy Practitioner, London
Dr Claire Anderson	Director of Pharmacy Practice & Social Pharmacy, University of Nottingham
Professor Elizabeth Anionwu	Professor of Nursing, Head of Mary Seacole Centre for Nursing Practice, Thames Valley University
Ms Audrey Ardern-Jones	Clinical Nurse Specialist, Cancer Genetics, Royal Marsden Hospital
Ms Glenda Augustine	Specialist Midwife, Haemoglobinopathies, W Midlands Perinatal Institute
Professor Mike Aulton	Professor of Pharmaceutical Technology, De Montford University, & RPSGB Representative
Mr Owen Barr	Lecturer, School of Nursing, University of Ulster
Dr Angela Barnicoat	Consultant Clinical Geneticist, London IDEAS Genetics Knowledge Park
Ms Carol Bates	Education Development Coordinator, Royal College of Midwives
Professor Sheila Bingham	Deputy Director, MRC Dunn Human Nutrition Unit, Cambridge
Ms Maureen Boxer	Lead Scientist, UK GTN
Mr Harper Brown	Chief Executive, Central Suffolk PCT
Mr Alan Burns	Chief Executive, Trent Health Authority
Mr Paul Chiles	Associate Director, Commissioning & Networks, Wandsworth PCT
Professor William Dawson	Managing Director, Bionet
Professor Anne de Looy	Head of the Department of Nutrition & Dietetics, Queen Margaret College, Edinburgh
Professor Stephen Denyer	Professor of Pharmacy & Head of School of Pharmacy, University of Brighton
Professor Dian Donnai	Professor & Director of the Department of Medical Genetics, St Mary's Hospital, Manchester
Dr Ruan Elliot	Nutritional Genomics Programme Leader, Institute of Food Research, Norwich Research Park
Dr Ian Ellis	Senior Lecturer in Clinical Genetics, Alder Hey Children's Hospital, Liverpool
Dr Jon Emery	Cancer Research UK Clinician Scientist, University of Cambridge
Dr Tony Farine	Specialist Subject Leader, Biological Sciences, School of Nursing, Nottingham
Professor Peter Farndon	Director, West Midlands Regional Genetics Centre, Birmingham Women's Hospital
Professor Steve Field	Regional Postgraduate Dean, University of Birmingham & West Midlands Deanery
Ms Sarah Forester	Professional Officer, Education, CPHVA
Dr Stephen Gillam	General Practitioner & Attachment Director, Department of Public Health & Primary Care, Cambridge University
Mr Jack Gillon	Acting Medical Director, National Services Division, Common Services Agency, Edinburgh
Dr Jonathan Gray	Clinical Director, Institute of Medical Genetics, University Hospital of Wales
Professor Neva Haites	Director, Clinical Genetics Service, Aberdeen University
Dr Rhydian Hapgood	MRC Fellow, University of Sheffield
Dr Hilary Harris	General Practitioner, GenEd, Manchester
Mr Chris Heginbotham	Chief Executive, Mental Health Act Commission, Nottingham
Ms Caroline Hurren	Head, Consultation & Education, Medicine in Society Programme, The Wellcome Trust
Dr Linda Hutchinson	Director of Education & Workforce Development
Mr Clive Jackson	Director, National Prescribing Centre, Liverpool
Dr Fred Kavalier	Primary Care Geneticist, Guys Hospital, London
Dr Steve Kelly	Director of Postgraduate General Practice Education, West

Mr Alastair Kent	Midlands Deanery
Dr Hossein Khaled	Director - Genetics Interest Group, London
Dr Maggie Kirk	Head of Health Improvement, Central Suffolk PCT
	Associate Head of School (Research) & Principal Lecturer, Genomics Policy Research Unit, University of Glamorgan
Ms Norma Lauder	Manager of the Department of Nutrition & Dietetics, Doncaster Royal Infirmary
Ms Janet Lawrence	Specialist Nurse, Haemoglobinopathies, Heart of Birmingham Teaching PCT
Professor Alain Li Wan Po	Professor of Clinical Pharmaceutics, Aston University
Ms Jacqueline Lowdon	Chief Paediatric Dietitian, University Hospital of Wales
Dr Anneke Lucassen	Senior Lecturer & Consultant in Clinical Genetics, Wessex Clinical Genetics Service
Dr James Mackay	Consultant Clinical Genetic Oncologist, NE Thames Clinical Genetic Service
Dr Alex Magee	Consultant Clinical Geneticist, Belfast City Hospital
Dr Richard Mann	General Practitioner & Trainer, Salisbury
Dr Alison Metcalfe	Research Fellow, School of Health Sciences, University of Birmingham
Professor Tony Moffat	Chief Scientist, Royal Pharmaceutical Society of Great Britain
Professor Patrick Morrison	Professor of Human Genetics & Consultant in Clinical Genetics, Belfast
Mr Albert Njindou	Regional Care Advisor, Huntington's Disease Association
Dr Maria O'Connell	Deputy Head of Micronutrient Status Research, MRC Human Nutrition Research, Cambridge
Ms Rose Marie Parr	Director of SCPPE & Lecturer, School of Pharmacy, University of Strathclyde
Dr Mary Porteous	Consultant Clinical Geneticist, Western General Hospital, Edinburgh
Ms Madeleine Rae	Cancer Network Manager, Humber & Yorkshire Coast Cancer Network
Dr Peter Rose	General Practitioner & Lecturer, Department of Primary Care, University of Oxford
Dr Christine Samson	Project Manager, Child Health Screening, UK National Screening Committee, London
Dr Tokunbo Sangowawa	Director of Public Health, North Tees Teaching PCT
Mr John Sargent	Chief Executive, Greater Manchester WDC
Dr Rosalind Skinner	Principal Medical Officer, Scottish Executive Health Department
Dr Heather Skirton	Nurse Consultant in Clinical Genetics, Taunton & Somerset NHS Trust
Ms Ann Smith	Principal Lecturer, School of Health, Northumbria University
Professor Peter Soothill	Professor of Maternal & Fetal Medicine, St Michael's Hospital, Bristol
Mr Mike Spencer	General Manager, Clinical Support Services, University Hospital of Wales
Dr Anna Stone	General Practice Registrar, West Midlands
Ms Michelle Styles	Head of Information Services, National Pharmaceutical Association, Herts
Ms Jane Thomas	Senior Lecturer, Department of Nutrition & Dietetics, Kings College, London
Ms Bernice Tighe	Senior Lecturer, Coventry University
Ms Sarah Wakefield	Research Fellow, CRMDE, University of Birmingham
Ms Lynne Watson	Specialist Health Visitor for Children with Special Needs, Twickenham & The Hampton PCT
Dr Douglas Wilcox	Senior Lecturer, University of Glasgow, Honorary Consultant in Medical Genetics & Director of Education, Yorkhill NHS Trust
Ms Rosie Wilkinson	RCN Nurse Advisor, Royal College of Nursing
Professor Joy Wingfield	Professor of The Pharmacy School, University of Nottingham
Ms Melissa Winter	Membership Engagement Officer, Genetic Interest Group, London
Ms Annie Young	Nurse Director - Three Counties Cancer Network

Project Team

Dr Hilary Burton

Consultant in Public Health Medicine, Public Health Genetics Unit, Cambridge

Professor Janet Grant

Director, Open University Centre for Education in Medicine

Mr Mike Greenwood

Media Analyst, London

Ms Denise Jillions

Public Involvement Manager, Public Health Genetics Unit, Cambridge

Ms Helen Jones

Project Researcher, Open University Centre for Education in Medicine

Dr Mark Kroese

Specialist Registrar in Public Health Medicine, Public Health Genetics Unit, Cambridge

Ms Heather Owen

Senior Research Officer, Open University

Dr Alison Stewart

Chief Knowledge Officer, Public Health Genetics Unit, Cambridge

Observers

Dr Sarah Bronsdon

Senior Project Manager, Medicine in Society Programme, The Wellcome Trust

Dr Alison Hill

Medical Advisor to the Department of Health & General Practitioner

Ms Dianne Kennard

Team Leader, NHS Genetics Team, Department of Health

Mr Roni Liyanage

Project Manager, Partnerships, The Wellcome Trust

Appendix 2: Method for stakeholder workshops

Workshop outline

The workshops were conducted in the following main phases:

- Presentation of background to the project and overall project structure
- Presentation of views from research including scientific about implications of genetics for that professional group
- Presentation of views from continuing education sector
- Presentation of public involvement paper
- An opportunity for each group member to describe their special viewpoint and to put forward any further concerns about issues that would have to be addressed.
- A prioritising exercise 'nominal group process' in two groups to look at clinical and teaching priorities
- A final review discussion

The nominal group process

The meeting was split into two groups and each was taken through a nominal group process in order to discuss and prioritise topics for education in genetics and the development of educational resources themselves. Group members were asked to spend a few minutes writing down their own thoughts in two areas:

- The priority areas for genetics in postgraduate medical education
- The priorities for the development of educational resources to provide this education

These views were then collated on a flip chart, discussed, clarified and overlapping statements rationalised by the group to create one coherent list. Group members then reviewed the lists separately and selected their five most important items from each list, allocating five points to the most important item and one to the least important of those five items. Participants wrote scores next to the items on the charts and then these were totalled, so that the group view of the relative importance of the topics on each list was obtained.

Rationalisation and comparison of prioritisation between groups

The topics mentioned by all the groups were rationalised as far as possible, and a final list generated. To compare groups and draw some general conclusions about priority topics, those topics that received votes for each stakeholder group were ranked. Groups voted for between 11 and 15 topics. To summarise, the top third rank of topics were given 3 stars, the middle third 2 stars and the lower third 1 star.

Appendix 3: Table of projected costs for a Centre for Genetics Education

Outline of likely costs associated with a Centre for Genetics Education based on experience from Cambridge Genetics Knowledge Park (at 03/04 price base):

Staff costs*	£thousands
Director	58
Project manager - medicine	52
Project manager - nursing	52
Project manager other professions	52
Professional adviser -education	52
Genetics adviser - education	32
Genetics adviser - communication	32
Information officer (database/librarian)	32
Communications officer	32
Media/website adviser	30
Business manager	32
Administrator (including support to Steering Group)	20
Events manager	24
Clerical officer/reception	14
Total staff costs	514
Total non-staff costs**	190
Total annual cost	703
Start-up***	100
*staff costs based on NHS pay scales. Employers on-costs are likely to be higher for the university contracts	
**based on costs of CGKP as comparable centre. This includes such elements as general office equipment, staff travel and subsistence, computer charges and maintenance, use of external contractors, overheads to employing organisation, network charges and maintenance, rent	
*** based on comparable set-up costs for CGKP	