Bridging the gap

PHG Foundation is an independent not-for-profit policy research organisation, working to improve human health by aiding the transfer of genome-based science and technologies from research into practice.

The Foundation bridges the gap between the development of new diagnostic and therapeutic tools in research, and their introduction as services to patients. We do this by:

- Providing knowledge, evidence and ideas to inform policy debate
- Engaging with policy makers and stakeholders to stimulate changes in public policy to support biomedical innovation and its application to improve health
- Helping health care providers and commissioners to evaluate the outputs of research, and to redesign services to deliver the most effective biomedical innovations to patients

What does ‘PHG’ stand for?

The Foundation grew out of the Public Health Genetics Unit, established with public funding in 1997 in Cambridge UK, one of the world’s most vibrant bioscience clusters. Ten years later, ‘PHGU’ has earned a national and international reputation as a leader in the field of public health genomics, with connections in the health, research and policy communities in the UK, Europe, North America, the Far East and Australasia. From these roots, we launched the Foundation as an international charity in May 2007, choosing to keep PHG in our name, both as a reflection of our heritage and to denote our focus on population- and public health and genomics.
Chairman’s foreword

Genomic science is in a robust state but there is widespread concern about the slow rate of progress in translating scientific advances into practical applications for diagnosing and treating disease and disability. Research funders have been increasing their calls for ‘translational research’ and we can expect to see significant growth in the number of potential new applications over the coming years.

However, the transition from promising new technology to application in mainstream health care is not a simple matter. Whilst the focus remains very much on the scientific research, this final stage of the translation process receives much less attention but is essential to realising the benefits of our huge investment in the basic science. Here, policy makers and health care providers have to evaluate new tests and interventions, make decisions about their utility and effectiveness, and plan for the impact on clinical practice. The interests of patients and their families, and the wider interests of our society as a whole, need to be considered. And we should be concerned to ensure that global health policy enables populations around the world to benefit from advances in genome-based science.

The complexity of the science on the one hand and the huge everyday demands on health services on the other, are creating a widening gap that needs to be bridged by a process of change management based on knowledge integration, knowledge brokering, stakeholder dialogue, education, and consensus-building. The team at the PHG Foundation have an outstanding track record in these areas and a deep understanding of the science, public health and the policy context. I was delighted to be asked to take on the role of trustee and chairman when the Foundation was launched in 2007, and I am very pleased to welcome you now to our first Annual Review, which I hope will serve as an introduction to PHG, its work and its people.

Professor Sir Brian Heap CBE, ScD, FRS
Creating the conditions for change

Dr Ron Zimmern, Executive Director, reviews our first year

Scientific research is giving us unprecedented new opportunities for treating and preventing disease that have significant implications for health care. The PHG Foundation’s aim is to create the conditions where policy makers and health care providers have the knowledge, motivation and resources to evaluate and deliver new technologies and medical interventions to patients. Our strategic priorities are contributing to better health policy and more effective health services to help our society realise the benefits of biomedical research and improve population health.

The Foundation’s policy work takes the form of both contributions to policy processes, and projects relating to particular needs and issues. One main strand this year has been tackling the lack of systematic evaluation and regulation of the burgeoning number of genome-based tests available for clinical use. Unlike medicines, there are no requirements for suppliers to provide evidence of their products’ clinical performance, despite the fact that the use of tests without proper evaluation may produce false results and lead to the wrong treatment being given.

We’ve been working with key stakeholders in the UK and in May we published a report on this issue, together with the Royal College of Pathologists. There are real signs that the Government is now seeing this area as a priority for further work. As a reflection of the global nature of markets for diagnostics, we are also working with colleagues in Europe and further afield to influence international policy and regulation. There is more about this in the review of our policy work on page 7.

Over the last ten years we have developed a great deal of expertise in defining the impact of new genome-based knowledge and technologies on individual specialties within mainstream medicine. This year we completed work on genetics in ophthalmology and we started two new projects on cardiac genetics and a new technology using cell-free fetal DNA and RNA for non-invasive prenatal genetic testing. Find out more on page 9. We use these projects to engage with policy makers and health care providers to improve services for patients. Next year we plan to bring together our experiences to create a generic model that health care providers can use for themselves as a tool for planning for the impact of genomic advances on their services.
I believe that one of the Foundation’s strengths is our ability to consider issues in depth and in the round, gathering evidence and bringing together insights from science, public health, clinical medicine, law and the social and population sciences with a practical perspective. We place the results of our work in the public domain for the use of researchers, health professionals, policy makers and others with an interest in biomedical innovation and health. We also share our knowledge with others through participation in education and other dissemination activities. See page 11 for some of this year’s outputs.

Looking ahead, our programme priorities will continue to focus on the policy and service implications of new research findings as they emerge. New projects for next year will include a review of the adequacy of consumer protection for genetic tests sold direct to consumers ‘over the counter’ and via the internet, and contributing to an EU-funded collaborative project on gene-environment interaction in ovarian, breast and prostate cancer. And in future I and my fellow trustees would like to see the Foundation explore ways to see that populations in developing countries get full benefit from genome-based science too.

Our corporate priorities for 2008/09 will be to ensure that we have the resources - money, strategic relationships, and high quality staff - to achieve the Foundation’s aims.

Full details of past and current work can be found on our website at www.phgfoundation.org

“Our strategic priorities are contributing to better health policy and more effective health services to help our society realise the benefits of biomedical research and improve population health.”
Developing ideas and influencing health policy

The Foundation’s policy programme looks at how national and international regulatory systems, the social environment and ethical debate interact with health needs, health care provision and the realisation of the benefits of scientific research. Some of the basic principles that underpin our approach to public policy are:

- We support the current pluralistic approach to policy-making but believe that, when it comes to genomic medicine, public health perspectives are under-represented.
- Public health thinking needs to move away from exclusive emphasis on the social and environmental causes of disease, towards a more sophisticated understanding of the interplay between genomic and environmental factors in disease causation.
- An overly complex legal environment is problematic for the development and translation of genomic medicine.
- Regulation should be confined to ensuring the safety of new tests and technologies, but manufacturers should be required to put evidence of the performance of their products into the public domain so that consumers can make informed decisions.
- There should be a better balance between individual privacy and the wider public good in the regulation of the use of personal data in research.

- ‘Genetic exceptionalism’, that is the belief that genomic information is uniquely powerful and requires special treatment, should be discouraged; DNA is just one of many types of biomarker, and other types of information may be equally or more predictive of disease risk.
- Both public and professionals need better understanding of the concepts of disease risk, and more research is needed on how people perceive and respond to disease risk information.

This year the Foundation’s research has focused on reviewing the extent of public European and international law as it relates to human biomedical science, and analysing its implications for policy development in the UK.

We have contributed to emerging legislative developments on data protection, human fertilisation and embryology, and health care provision. In the spring we were called to give evidence to the House of Lords Science and Technology Committee’s inquiry into genomic medicine.

However, the evaluation and regulation of diagnostic tests has represented the main theme for our multidisciplinary work.
Research is producing lots of new information about possible links between genes and disease, giving rise to novel tests for diagnosis and prognosis, as well as for tolerance to medicines. Alongside the use of new tests in clinical medicine, the commercial market for direct-to-consumer tests is growing, with claims of being able to predict the likelihood of future disease for individual users.

Despite the increased use of genetic tests, there is no formal evaluation system to ensure they are effective or useful. Nor is there any requirement for test suppliers to provide evidence of a test’s clinical performance. Whilst the tests themselves are rarely harmful, the results gained from unvalidated tests could lead to the wrong treatment being given, or false reassurance that prevents further treatment or causes individuals to ignore effective prevention measures. Pressure is now mounting for a formal evaluation process for genetic tests, and indeed for all clinical biomarker tests, but no consensus has been reached about how best to achieve this.

The PHG Foundation has taken a lead in analysing the conceptual issues involved, developing a coherent evaluation framework, and driving national and international policy through examination of the possible regulatory hurdles and solutions.

In January 2008 we co-hosted, with the Royal College of Pathologists, an expert ‘diagnostics summit’ to discuss the evaluation and regulation of laboratory tests in use within the NHS. The summit called for the establishment of a new body to ensure the evaluation of diagnostic tests, and the creation of a publicly available database of new and existing laboratory tests – a ‘diagnostics formulary’ – containing evidence of clinical performance or indeed explicit reference to the lack of it. The recommendations were presented to MPs at a parliamentary briefing in May, and are currently being discussed within the NHS.

In the spring of 2008, we presented our work to the Committee on Safety of In Vitro Devices, part of the Medicines and Healthcare products Regulatory Agency (MHRA). Later this year we will also be working with the International Organisation for Economic Cooperation and Development (OECD), which helps governments tackle the economic, social and governance challenges of a globalised economy, to deliver a workshop on policy issues for the development and use of biomarkers in health.

Next year we will turn our attention to ‘genetic susceptibility’ tests, that is, those used to predict the likelihood of developing a disease later in life, and their potential use for population screening. Current evaluation models may not be suitable for these predictive tests, and we will be doing some methodological work to assess the adaptations that will be needed. Alongside this we will look at the growing market for predictive tests as a ‘lifestyle’ product, and consider whether consumers are adequately protected by current legislation.

**Focusing on safeguards for new diagnostic tests**
The main objectives for future development of the Foundation’s policy work are twofold. Our primary goal is to begin to broaden the clinically-oriented translational research of the Foundation by facilitating a greater understanding of the complex technical, political, legal and ethical context in which the management of biotechnologies is undertaken. Secondly, we want to intensify our dialogue with external colleagues, policymakers and other stakeholders, to ensure that the work of the Foundation becomes more widely known to decision-makers at all levels in national and international health policy processes.

Inherently, our policy work has a strong analytical and academic dimension, and the Foundation plays an important enabling role by fostering a robust intellectual environment for the dynamic interchange of ideas within and across disciplines. The Foundation has already made a long term commitment to fund three college lectureships - in law, philosophy and social science - at the new Centre for Biomedical Science in Society, Hughes Hall, Cambridge, established this year under the directorship of leading health psychologist Professor Theresa Marteau.

In addition to teaching and research, these posts will contribute academic content to Foundation projects through the involvement of the lecturers as participating members of our expert working groups.

We also plan to strengthen the Foundation’s community of academic and professional support by more formally recognising our relationships with ‘senior faculty’ - leading experts and commentators from a range of disciplines - whom we invite to work with us on strategic projects. Further, capacity-building within the policy team will enable us to take on new projects and intensify our research across the work programme.

Our work on the regulation of diagnostic tests and biomarkers is likely to be completed by the end of 2008, and with expanded capacity we will be able to add at least one further project centred on legal and philosophical issues which impact on the support and regulation of health innovation and technology. We are currently considering the implications of affordable genome sequencing for personalised medicine, as well as governance of the use of tissue, data and information to facilitate long-term, large sample, international research.

Dr Stephen John
PHG Foundation Lecturer in Philosophy, Hughes Hall Cambridge
Applying knowledge and ideas to change health services

The Foundation aims to identify the potential implications of emerging genome-based science and technologies, and to engage with policy makers and health care providers to develop services and public health programmes that will deliver benefits to patients and their families.

This year we presented a report on genetics in ophthalmology to the UK National Health Service, detailing the requirement for specialised services for people with inherited visual impairment. It draws attention to the need for health services to anticipate extra demand arising from the expected growth in the number of genome-based tests and treatments available, not just for relatively rare conditions but also for the common eye diseases such as age-related macular degeneration and glaucoma. We have been asked to adapt this work into guidance for service commissioners and local service providers; and to inform patients about what they can expect from the NHS in terms of effective, high quality care.

Our work on the potential use of a new technique for non-invasive prenatal genetic testing, using cell-free fetal nucleic acids in the maternal blood, is attracting a lot of interest. The possible introduction of this technology into routine antenatal screening raises significant ethical and logistical challenges, and we are working with experts and stakeholders to evaluate the use of the technology for different applications. We will publish a strategy for its implementation in spring 2009.

We also began a major two-year project on cardiac genetics services and the prevention of sudden cardiac death in the UK. We are studying the epidemiology of single gene disorders related to heart disease and the state of the clinical science for diagnosis and treatment of these disorders. We are considering the ethical and legal issues, particularly those surrounding the sharing of genetic data and the management of human tissue samples. We will be looking at current service provision and reviewing the relationship between specialist genetics and cardiac services. We will also be quantifying current and future demand for these services and listening to patients and their families who are affected by these conditions. The work will culminate with a report of our findings and recommendations for the key components of cardiac genetics services and related elements in primary and secondary care, pathology and coroner services.

The UK’s National Screening Committee asked us to coordinate a review of its 10 year-old carrier screening programme for Tay Sachs Disease, an inherited degenerative neurological disease that particularly affects the Ashkenazi Jewish population. This work is being undertaken with researchers from the genetics team at Guy’s Hospital, London, and findings will be published in autumn 2008.
Dr Hilary Burton, Programme Director, sets out plans for next year

Our completed projects on how emerging scientific discoveries impact on mainstream health services have a number of common elements:

- a review of the state of the science
- an estimate of the potential demand for services based on a health needs assessment
- a review of the current service provision
- service user involvement
- a look at how the introduction of genome-based elements to services will affect the volume and nature of the work
- identification of potential barriers to progress and policy option proposals for future development of services

Next year I want to bring these elements together into a generic service development model and explore the potential for its use by health care providers for their own service planning.

I am also looking forward to our involvement with a new EU-funded study that starts in 2009, to find ways to identify individuals with an increased risk of breast, ovarian and prostate cancer. We will be working alongside some of the most eminent researchers in Europe, Scandinavia and Australia on this four-year project that will lead to improved understanding of the genetic and environmental basis of carcinogenesis and the development and use of new risk prediction tools.

On the horizon, epigenetics, the study of changes in gene function that pass from parent to child without a change in DNA sequence, is a fascinating area of science with the potential for impacting on health care. We will be looking at the published research and talking to leading experts to assess what the implications might be and whether the time has come for thinking in depth about the policy and service issues.

One of the Foundation’s strengths is our in-house capacity for epidemiologic reviews of the incidence and distribution of disease in the population as a key part of our service development work. The Foundation has awarded a two-year grant to the Medical Research Council's Cambridge-based Biostatistics Unit to fund their contribution of epidemiology skills to our work programme. The grant also enables their staff to provide the UK Coordinating Centre for the International Human Genome Epidemiology Network (HuGENet).
Sharing knowledge and ideas

Knowledge is at the core of what we do. In the course of our work programme, we acquire a lot of it: from studying the outputs of research (biomedical science, population sciences and the humanities), and from listening to the experiences of health and policy professionals, and those of users of health services and their families.

We use knowledge in two ways to influence health policy and service delivery. Firstly, we synthesise knowledge from different areas of expertise to create actionable ideas and solutions to health needs and translational challenges. And secondly, we ensure that our change management activities are underpinned with expert knowledge and research-based evidence.

Our projects provide spaces where practitioners, policy makers, researchers, and consumers, are brought together to enrich the research and decision making processes.

Outside of our project work, we support researchers and health professionals through dissemination of knowledge in other ways: via our online news service (including comment and analysis), information resources, publications, briefings, education activities and events.

This year we took part in a number of national and international health policy networks and Foundation staff served on committees and working groups for a range of influential public service and non-governmental organisations. The Foundation provided a formal advisory function to the Chief Medical Officer of the Welsh Assembly Government and to EuroGentest’s ‘Capability’ project.

Our education activities have included contributions to textbooks on public health and human genome epidemiology, teaching on undergraduate and postgraduate courses and leading work on education strategies for the European PHGEN network. We were commissioned to provide a three day examined course in genomics and health policy for the University of Hong Kong in November 2007, and provided a three-month training placement for an official from the Hong Kong Department of Health at our Cambridge base.

We are accredited to provide training places for public health trainees, and we always welcome enquiries from visiting scholars who want to spend time with us.

Public health genomics can be fun - PHG Foundation at Cambridge Science Festival 2008
This year:

- Our website received 103,000 visits from 180 countries/territories
- Our genomics policy news pages were the most popular (50% of visits)
- The number of people signed up to receive our online newsletter doubled to almost 900
- We had almost 800 new registrations for our online information resources
The information given here is taken from the statutory accounts for the year ended 31 March 2008, on which our auditors have given an unqualified report, and which have been lodged with the Charity Commission and Companies House. Please refer to the statutory accounts for a full understanding of the Foundation’s financial affairs. These are available on our website or we will supply a copy on request.
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Julie Towers - Finance Manager
Joanna Trautsolt - Administrator

**Development and Fundraising**

Shelley Gregory-Jones - Development Director
Conrad Chua, Nicola Longdon - Development Associates
The Trustees and staff would like to thank our funders and supporters for their donations, grants, advice and contributions to our projects this year. Particular thanks go to Alan Barrell for his help with our development programme, and to Sarah Squire for her enthusiasm and support in establishing our relationship with Hughes Hall.

Our two corporate priorities for 2008/09 are:

• **Raising money** to continue our work beyond 2012, and
• **Strategic partnerships** to bridge the gap between scientific research and health care policy and practice

If you would like to get involved, please contact Shelley Gregory-Jones at the address below or telephone 01223 740328.