Our vision for the 21st century is to see the benefits of society’s investment in scientific and medical research realised as better health and wellbeing for people around the world.
PHG Foundation is an independent, not-for-profit public health organisation with the mission to mobilise innovation in biomedical science to improve the health of populations worldwide.

Scientific research is creating unprecedented opportunities for diagnosis, prevention and treatment of illness and disability. PHG Foundation tackles some of the real gaps and barriers that hamper the translation of new technologies from research into practice and adoption by health systems.

We do this by:

- Engaging at a strategic level with UK and international stakeholders to address policy gaps, ethical concerns and regulatory challenges
- Helping healthcare organisations to redesign their programmes and services to deliver effective biomedical innovations to improve the lives of people in developed and developing nations, with a focus on the most vulnerable
- Providing knowledge, evidence and ideas to inform translational research, policy discourse and public debate

Our programmes and projects are described more fully on our website at www.phgfoundation.org
Welcome to this review of our work and achievements for 2009.

The current recession is focusing everyone’s minds on cutting costs and getting the best possible value for money; not least in biomedical research where the spotlight is on translation and the development of applications with practical benefit for patients.

The Foundation’s programmes are showing, in a practical way, how we can evaluate new technologies, improve investment decisions, and develop our health systems to deliver better care for patients and their families. But in a broader sense, the Foundation is also looking at how health care systems need to adapt, not just to the possibilities of 21st century biological science, but also to changes in society and the environment. The trend towards individual autonomy and choice, the empowerment of individuals through electronic access to information, the potential offered by genomics for so-called ‘personalised medicine’ and the growth in consumerism all present huge challenges to the collective concepts of public health and the state’s role in protecting the health and wellbeing of its citizens. At a policy level, the Foundation is fostering debate about how we deal with these challenges and at the same time protect our traditional public health objectives of providing population-based health care that is effective, evidence-based, inclusive and accessible to all based on need.

Such public health values are universal and not just the province of high-income, developed nations. In health matters, we are a global community, as the recent swine flu pandemic has clearly demonstrated. The Foundation is taking the debate about 21st century science and the future of public health to international policy makers and opinion formers, not least to add to the momentum for ensuring that low- and middle-income countries also benefit from biomedical innovation. Our own programmes are starting to look for practical ways of harnessing new genome-based knowledge and technologies to improve health outcomes for those most in need in developing nations, and offering them tools to develop services that are sustainable, effective and suited to their local circumstances.

The international agenda is a top priority for our new Chief Executive, Dr Mukesh Kapila, who joined us in September. Mukesh is a public health physician, best known for his work in international development and humanitarian affairs, and is passionate about liberating people from the effects of illness and poverty. These concerns are, I know, also close to the heart of my predecessor as Chairman, Sir Brian Heap, who has stepped down after seeing the organisation through its first two years. We are immensely grateful for his work and delighted that he continues his involvement with the Foundation as a trustee. I am proud to have the support of my fellow trustees and the fine team of staff at the Foundation who, as I hope you will see from this review, share the belief that a strong public health approach is crucial to bridge the gap between the development of new technologies and their application to bring health for all.

Ron Zimmern
Chair of Trustees
September 2009
PHG Foundation engages in the translation process by using organised, multidisciplinary action to link the ‘culture of enquiry’ in biomedical research with those whose job it is to deliver health policy and patient care.

Policy makers and research funders are calling for more “translational research”, first to turn basic scientific discoveries into new products or interventions, and second, to transfer these new products into everyday health care. However, thinking of translation just in terms of research, which is essentially a culture of enquiry, does not go far enough.

Organised, cross-sector action is needed to create the conditions in which health care systems can adopt biomedical innovations and build them into effective services to patients. With our public health grounding, PHG Foundation works in this area, conducting demonstration projects and advocating for more attention to be paid to bridging the gap that exists between translational research and the adoption and diffusion of innovations into healthcare practice.

The Foundation’s work on inherited cardiac conditions was recognised with the ERBI NHS Partnership Award for 2009.
Common chronic diseases: Inherited cardiovascular disease

Inherited cardiovascular conditions are a group of genetic disorders that affect around 340,000 people in the UK. Sometimes, the first indication of a problem is the sudden and unexpected death of an apparently healthy young person. Obviously, other relatives may also be at risk. With recent advances in genetics and cardiology it is now possible to reduce the number of tragic and unnecessary deaths by offering much more accurate diagnosis, improved clinical management, and better risk assessment for affected families. But such services are not available on a consistent basis in the UK.

We brought together a group of experts, service providers and patient support groups to survey the state of the science, carry out a needs assessment for cardiac genetics services to care for affected families and review current service provision. We found evidence of serious inequalities in both quality and availability of services across the UK; existing services are insufficient to meet current needs, let alone to meet the anticipated growth in demand. The project’s findings and recommendations for improving health care for affected families have been warmly received by the Department of Health. As follow-up, the Foundation has been commissioned to work with a small group led by the NHS Heart Improvement Programme, to develop guidance for commissioners.

Maternal and child health: Prenatal testing using cell free fetal DNA

Over a decade ago, scientists made the landmark discovery that fetal DNA is present in maternal blood during pregnancy. The fact that small quantities of this so-called ‘cell-free fetal DNA’ can be reliably detected from a sample of the mother’s blood just 7 weeks after conception offers important new opportunities for earlier diagnosis of genetic conditions, without the risks and costs associated with the current, invasive methods of prenatal testing. But to date, roll-out of this new technology as a widely available test in public health services has not been achieved.

There are also some important ethical questions, not just associated with clinical use of the technology but also with its non-medical applications, for example sex selection for social reasons and paternity testing. These need to be addressed, particularly as the test is becoming increasingly available direct to consumers over the internet, without professional counselling.

PHG Foundation brought together technical experts, NHS service providers and wider stakeholders (including patient groups) to develop a strategy for evaluation and funding delivery of this new technology to patients across the UK. As a result of this work, the National Screening Committee has issued a fact sheet for the public and PHG Foundation is taking part in a large national research collaboration to develop this technology for various applications in the UK.

Inherited cardiovascular conditions are a group of genetic disorders that affect around 340,000 people in the UK. Sometimes, the first indication of a problem is the sudden and unexpected death of an apparently healthy young person. Obviously, other relatives may also be at risk. With recent advances in genetics and cardiology it is now possible to reduce the number of tragic and unnecessary deaths by offering much more accurate diagnosis, improved clinical management, and better risk assessment for affected families. But such services are not available on a consistent basis in the UK.

We brought together a group of experts, service providers and patient support groups to survey the state of the science, carry out a needs assessment for cardiac genetics services to care for affected families and review current service provision. We found evidence of serious inequalities in both quality and availability of services across the UK; existing services are insufficient to meet current needs, let alone to meet the anticipated growth in demand. The project’s findings and recommendations for improving health care for affected families have been warmly received by the Department of Health. As follow-up, the Foundation has been commissioned to work with a small group led by the NHS Heart Improvement Programme, to develop guidance for commissioners.

When an inherited cardiac condition is diagnosed or suspected, health professionals often face real tensions between their duty to protect the privacy and rights of the affected individual and their obligations to the wider family, who also have an interest in gaining access to effective diagnosis and treatment. We are calling for legal and regulatory action to ensure appropriate retention of tissue samples following a sudden cardiac death; to improve care for at-risk relatives; and to promote best practice in consent processes and data sharing.

See our report: Cell-free nucleic acids for non-invasive prenatal diagnosis, January 2009

See our report: Heart to Heart: Inherited Cardiovascular Conditions Services, June 2009
PHG Foundation engages in the policy process to strengthen health systems to deliver biomedical innovation in ways that are efficient, effective, equitable and sustainable.

Health systems in the developed world are under pressure from the “push” of innovative technologies and the “pull” of increased demand fuelled by new diagnostics and predictive tests. They face fundamental challenges in containing costs and reconfiguring their services to deliver new technologies.

As a society we have a moral imperative to ensure that people in low and middle income countries also benefit from scientific advances, and we must work towards the establishment of health systems in the developing world that provide access to modern genetics services that are appropriate and affordable in their contexts.

PHG Foundation’s programmes address some of the horizontal policy issues and responses to such challenges faced by health systems in the UK, Europe and internationally.
Genetic testing and biomarkers

There has been a massive expansion in research into the contribution of genetic factors to complex multifactorial diseases such as cancer and heart disease. Reliable evidence of associations between genetic variants and the risk of developing specific diseases is rapidly emerging, giving rise to many new tests and biomarkers that can be used to determine the presence of a disease, predict the risk of developing a disease in the future, or to predict an individual’s reaction to drug treatment.

The commercial market is burgeoning with direct-to-consumer tests that claim to predict the likelihood of onset of disease later in life. The use of such tests without proper evaluation or support in interpreting the results can be harmful, perhaps prompting inappropriate treatment based on an unreliable estimate of risk or inaction based on false reassurance of low risk.

PHG Foundation has been at the forefront of advocating for a system of formal evaluation of new genetic tests and biomarkers, and has been influential in the development of UK and European ethical and legal frameworks that govern the use of such tests in clinical practice and research. This year Ron Zimmern was invited to give the prestigious Milroy Lecture on this topic at the Royal College of Physicians in London. In September we ran a workshop for the Organisation for Economic Cooperation and Development (OECD) on Policy Issues for the Development and Use of Biomarkers in Health. Briefing papers and a report of the meeting are available on the OECD website and will form the basis of an OECD publication.

Our work in this area is now evolving towards the evaluation of risk prediction models and their use to determine thresholds for intervention in health systems.

Further details on our website at www.phgfoundation.org/pages/work7.htm

Maternal and child health: Improving care and reducing birth defects in developing countries

Better health for mothers and children is at the heart of the Millennium Development Goals. While we have seen significant reductions in childhood illness and deaths from infectious diseases in developing countries, the same cannot be said for birth defects, which are now associated with a third of deaths in children under five. Worldwide over 8 million children are born with serious birth defects of genetic or part-genetic origin, and another million are born with problems arising from exposure to environmental factors during pregnancy – most in developing countries. Beyond bare statistics, affected children and their families suffer not just the physical and emotional burdens but also stigma, discrimination and impoverishment.

But there is huge potential for reducing the burden of birth defects through better diagnosis, care and treatment. Practical improvements in basic services can help low-income countries, including through the development of cost-effective medical genetics services.

PHG Foundation is leading a multi-partner international collaboration to address these challenges. In Spring 2009 we began a five-year programme to develop a framework and toolkit that low- and middle-income countries can use to assess their care needs in relation to birth defects; and to develop services to deliver simple, cost-effective technologies and interventions that will improve maternal and child health. We are currently seeking additional funding partners to help support the first two phases of the programme which will develop the toolkit and pilot its application in at least two developing countries.

Further details on our website at www.phgfoundation.org/pages/maternal-and-child-health.htm
PHG Foundation looks at the future to consider the potential impact of scientific developments on public health and civil society, to contribute new ideas and evidence to the policy discourse.

Our growing knowledge of genomics, molecular and cell biology has structural implications for both our health systems and the way we think about the practice of medicine. Excitement about the opportunities offered by new technology must be tempered with solutions for new ethical and legal dilemmas. It is important to understand and influence the complex backdrop of political, economic, social and cultural factors that provide the context in which the translation process takes place if we are to avoid creating unnecessary barriers to realising the benefits for human health.

PHG Foundation’s approach is grounded in the realities of the present, but our Future Health programme looks at emerging trends in biomedical research and their wider implications for health and society. We contribute evidence and ideas to the global public health industry, focusing on practical applications of science and solutions to difficult policy problems. We act as a multiprofessional, independent voice to serve the public good by bridging the research-policy-implementation gaps; and by enriching the quality of debate and decision making through engagement, education and outreach activities.

Science and public health horizons

At PHG Foundation we think of biomedical innovation not just in terms of new diagnostic technologies, medical devices, drugs and clinical interventions; though undoubtedly this is where the ‘quick wins’ can be had if we can overcome some of the barriers to translation. We are also interested in how these emerging tools help or require us to think differently about the way we do things.

For example we are starting to see a wave of models emerging from research that aim to predict an individual’s risk of disease before symptoms appear or at some point in the future. The use of such “susceptibility” models has significant impact for clinical medicine, for public health in terms of novel methods for stratifying populations and targeting screening and other programmes, and for commercial suppliers of direct-to-consumer testing. There is an increasingly pressing need to develop standards against which these models can be judged, so that researchers, physicians, policy makers and the public can assess their validity and utility; and society can consider the wider implications of predicting future disease in this way. This year PHG Foundation began a series of linked projects that will develop a set of criteria for evaluation of risk prediction models; question some of the philosophical concepts that currently underpin thinking about genomics, epidemiology and risk; and develop a rational response to ethical and legal concerns about genetic discrimination.

PHG Foundation monitors and comments on proposed changes in policy, regulation and legislation and the potential impact on research and health services. This year we made contributions to the Coroners’ and Justice Bill, the Human Fertilisation and Embryology Bill, the Health and Social Care Bill, the House of Lords Science and Technology Committee proceedings and developments in EU legislation on data protection.

The practice of public health is another area that requires fresh thinking in the post-genomic era. It is not just that public health should grasp the new opportunities presented by modern science alongside its more traditional concerns of the social and environmental determinants of health. The profession is facing fundamental challenges to its core population-orientated, collectivist values and concerns for equity: our changing social culture emphasises individual autonomy and choice, information technology empowers the ‘expert patient’ and science offers the prospect of ‘personalised’ medicine.

PHG Foundation is opening the debate to consider how 21st century science will impact on the international public health movement and on the state’s role in protecting the health of its citizens.

Analysis, comment and advocacy

We responded to consultations run by the Human Tissue Authority, the General Medical Council, the NHS and the Association of British Insurers. Foundation staff serve as advisers on many national and international working groups and committees, for example in the NHS, and with the British Society for Human Genetics, Genetic Interest Group and the international Capability Project.
Chief Executive’s outlook

I am honoured to join such a talented and committed team at the PHG Foundation, brought together by the creative enterprise of Ron Zimmern and his fellow trustees. Their inspiration and guidance has already secured the Foundation’s position as a respected leader in public health genomics. I look forward to building on this to enable the Foundation to do yet more and reach even further.

I am often asked why I have accepted a role that appears, at first glance, to be somewhat removed from a well-established and all-consuming career in international diplomacy, development and humanitarian affairs. The answer is straightforward. Accompanying the pilgrimage back to my Cambridge professional roots is an intense impatience with business-as-usual. This is born out of over two decades of wandering around the globe that have brought me face to face with the repeated tragedies of an increasingly distressed world. Every day we surpass our own ingenuity in discovering more and more “wondrous things” but, every day, we fail in urgency and dedication to bring their benefits to the most desperate and vulnerable on our shared planet.

PHG Foundation is concerned with bridging this gap. Of course, we need to do more on many, many things. But more than that, we also need new, smart ways to address the myriad ills of our world. This is a world in which the forces of globalisation mean that the traditional division between rich and poor countries is no longer adequate to describe our inter-dependence, nor to define the inter-connectedness of challenges facing us and the unprecedented levels of co-operation needed to address them. Nowhere is this truer than in the enjoyment of health that is simultaneously a personal human right and a collective human good. Ultimately, this is about creating the security that people everywhere sorely lack, in a world of uncertainty and ambiguity.

PHG Foundation contributes to this global security through its emphasis on policy making that is evidence-based and population-centred, and by advocating and designing systems and capacities that will share the fruits of science and technology on an equitable basis.

In making science work for health, there are three crucial considerations for our forward thinking. First, how can we do better in educating the public and policy-makers so that there is well-informed debate on the potential and pitfalls of developments in the biosciences, leading to the adoption of sensitively-made and sensible choices? Second, how can we argue more strongly to orient investments in biomedical innovation and research towards the needs that matter most from the public health and social welfare perspective, in both low and higher income countries? Third, how can we accelerate the translation of biomedical innovation into life-saving and life-enhancing practices, ensuring that the poorest and most vulnerable - worldwide - are at the front of the queue to benefit?

These are my initial and personal reflections as I learn more from expert colleagues. We have initiated a “Vision 2020” process and invite all the stakeholders and friends of the Foundation to join us in shaping our strategy for the coming decade.

Dr Kapila joined the PHG Foundation in September 2009 after an initial career in the UK National Health Service and subsequently with the UK Department for International Development, followed by senior appointments at the United Nations and with the World Health Organization and the International Federation of Red Cross and Red Crescent Societies.
Financial information

The information given here is taken from the statutory accounts for the year ended 31 March 2009, on which our auditors have given an unqualified report. Please refer to the statutory accounts for a full understanding of the Foundation’s financial affairs. They are available online from the Charity Commission and Companies House, and can be downloaded as a pdf from the Foundation’s website.

<table>
<thead>
<tr>
<th></th>
<th>£ 000’s</th>
</tr>
</thead>
<tbody>
<tr>
<td>Donations</td>
<td>1,097</td>
</tr>
<tr>
<td>Grants</td>
<td>35</td>
</tr>
<tr>
<td>Investment income</td>
<td>82</td>
</tr>
<tr>
<td>Other</td>
<td>7</td>
</tr>
<tr>
<td><strong>Total</strong></td>
<td><strong>1,221</strong></td>
</tr>
</tbody>
</table>

2008/2009

Resources expended

<table>
<thead>
<tr>
<th></th>
<th>£ 000’s</th>
</tr>
</thead>
<tbody>
<tr>
<td>Programme costs</td>
<td>926</td>
</tr>
<tr>
<td>Fundraising</td>
<td>93</td>
</tr>
<tr>
<td>Governance and investment management</td>
<td>30</td>
</tr>
<tr>
<td>Balance (reserves)</td>
<td>172</td>
</tr>
<tr>
<td><strong>Total</strong></td>
<td><strong>1,221</strong></td>
</tr>
</tbody>
</table>

Grants given

We may make grants to organisations to partner with us to provide knowledge, services and infrastructure that support our strategic objectives. Application is by invitation only to organisations that we know well and trust. We do not respond to unsolicited applications and we do not normally make grants to individuals.

This year the Foundation awarded grants to:

- **MRC Biostatistics Unit**, to provide us with access to world-class statistics and epidemiology skills for our needs assessment and service review projects
- **Hughes Hall, Cambridge**, for three academic posts in law, philosophy and social science to give academic underpinning to the Foundation’s policy work and provide a base for raising further academic research grants through the Hughes Hall Centre for Biomedical Science and Society

In addition the Foundation has a limited small grants scheme that has supported collaborations with small charities and not-for-profit networks that share our aims.

Website

This year:

- Our website received 124,152 visits from 193 countries/territories
- Our Genomics Policy News pages were the most popular (54% of visits)
- 585 people signed up to receive our online newsletter (making a distribution list of over 1200 in total)
- We had more than 800 new registrations for our online information resources
Reports

Heart to Heart - Inherited cardiovascular conditions services: A needs assessment and service review

Tay Sachs Disease carrier screening in the Ashkenazi Jewish population: A needs assessment and review of current services

Cell-free fetal nucleic acids for non-invasive prenatal diagnosis: Report of the UK expert working group
Wright C. PHG Foundation (2009)

Ethical, legal and social issues arising from cell-free fetal DNA technologies
Hall A, Bostanci A, John S. Appendix III to C Wright’s report. PHG Foundation (2009)

Full details available on our website at www.phgfoundation.org/reports

Publications in academic journals

Policy papers


Scientific papers


Wright CF, Chitty LS. Cell-free fetal DNA and RNA in maternal blood: implications for safe antenatal testing. British Medical Journal 2009 339: 161-164


Full details available on our website at www.phgfoundation.org/papers

Books

• Hall A. Accessing genetic information: Anomalies arising from the Regulation of Genetic Material and Genetic Information in the UK. Invited book chapter as part of the ProEur Project. (to be published by Ashgate. Submitted February 2009)

• Zimmern R & Brice P. The Public Health Genomics Enterprise in Human Genome Epidemiology, 2nd edition (OUP), Ed. Muin Khoury


Lectures and presentations

Foundation staff were invited to speak at over 65 events this year. Highlights:

2008

• Ron Zimmern. 7 May: Milroy Lecture “Testing Challenges: The Evaluation of Novel Diagnostics and Molecular Biomarkers”, Royal College of Physicians, London

• Hilary Burton. 13 May: Presented to the Joint Committee on Medical Genetics: Genetic Ophthalmology in Focus

• Ron Zimmern. 1 June: European Society of Human Genetics. The Clinical Use of Genetic and Molecular Biomarkers: A Public Health Perspective

• Ron Zimmern. 25 June: Evidence to Select Committee on Genomic Medicine, House of Lords, London

• Hilary Burton. 6 October: Keynote speech to GIG at their annual conference on Choices & Challenges of Reproductive Health, Cavendish Square, London: Inherited Conditions: the Challenge for Health Services

• Caroline Wright. 23 October: Presentation to the Joint UK National Screening Committee and Human Genetics Commission workshop, London, on FFDNA

• Ron Zimmern. 6-7 October: OECD meeting, Hinxton, Cambridge. Evaluation and Regulation of Biomarkers: A Public Health Perspective

2009

• Ron Zimmern. 27 January: Manchester NOWGEN. The Evaluation and Regulation of Genetic and Molecular Biomarkers


• Philippa Brice. 2-6 May: Presentation at Genes for Health Conference, Perth, Australia entitled Towards implementation of non-invasive prenatal diagnosis in the UK National Health Service

• Hilary Burton. 2-6 May: Presentation at Genes for Health Conference, Perth, Australia entitled Predictive & susceptibility test evaluation – a public health perspective. Also Welcome GRaPH-Int and Closing Address.
Trustees

Professor Sir Brian Heap CBE ScD FRS
Baroness Onora O’Neill CBE
Ian Peacock MA
Sir Keith Peters FRS FMedSci
Sarah Squire MA (appointed Dec 2008)
Dr Ron Zimmern MA FRCP FFPFM

Institutional Members

Cambridge University Hospitals NHS Foundation Trust
NHS East of England
Hughes Hall, Cambridge
University of Cambridge Faculty of Law
University of Cambridge School of Clinical Medicine
University of Cambridge Department of History and Philosophy of Science
University of Cambridge Centre for Science and Policy

Staff

Management team
Chief Executive - Dr Mukesh Kapila (from Sep 2009)
Operations Director - Carol Lyon
 Programme Director - Dr Hilary Burton
 Head of Science - Dr Caroline Wright
 Head of Communications - Dr Philippa Brice
 Development Manager - Shelley Gregory-Jones

Programme staff
Project Manager - Corinna Alberg
Project Manager (Law) - Alison Hall
Consultant in Public Health - Dr Luis Nacul
Epidemiologist - Dr Gurdeep Sagoo
Project Coordinator - Dr Sowmiya Moorthie
Project Coordinator - Dr Susmita Chowdhury

Business support staff
Administration Manager / PA - Nicola Harvey
Administrator - Anke Friedrich
Finance Manager - Julie Towers
PA/Administrator - Jane Lane
Systems Developer - Calvin Cheah
Webmaster and Information Manager - Simon Leese

Programme associates

Principal Associate - Dr Mark Kroese
Principal Associate - Dr Simon Sanderson
Principal Associate - Dr Alison Stewart
Principal Associate - Dr Tom Dent
Principal Associate - Dr Layla Al-Jader
Hughes Hall lecturer (Law) - Dr Amit Pundik
Hughes Hall lecturer (Philosophy) - Dr Stephen John
Hughes Hall lecturer (Social Science) - Dr Adam Bostanci

Senior Fellows

Professor Arnold L Christianson
Dr John A Crolla
Professor Peter Farndon
Professor Steve Humphries
Mr Alastair Kent
Dr Christine Patch
Dr Paul Pharoah

Acknowledgements

The Foundation’s trustees and staff would like to thank our funders, associates and supporters for their donations, grants, advice and contributions to our projects this year.

If you would like to get involved in one of our programmes or make a donation to support our future work, please contact us at contact@phgfoundation.org

Donations can also be made online at www.phgfoundation.org/donate