Personalised healthcare: making the most of genomics

Tuesday 29 November
Crausaz Wordsworth Building, Robinson College, Cambridge
Welcome

It is a great pleasure to welcome such a rich variety of delegates to our latest Life Sciences & Society event. There can be no doubt that the breadth and depth of experience you bring will enhance and enliven our examination of the potential of genomics to deliver more personalised healthcare now and in the future – and the wider implications of these applications.

Today we take the opportunity to learn about selected uses of genomics for health (beyond the usual suspects of personalised cancer therapeutics and rare disease diagnoses) and consider practical and policy issues in greater depth. The final discussion will be around societal issues related to the use of genomics in healthcare, after which we hope you will stay and continue the discussion over drinks.

Life Sciences & Society series

Healthcare is on the cusp of an exciting new future, but health systems must move quickly to meet these challenges and exploit the opportunities ahead. The Life Sciences & Society series is a range of events that highlight key areas where science and technology could transform disease prevention and care, examine barriers to implementation, and explore the policy implications of biomedical and digital innovations for health.

Events are open to all and combine expert insight with lively discussion and networking. For more information, go to: www.phgfoundation.org/LSSS

PHG Foundation
Making science work for health

The PHG Foundation is a health policy think tank with a special focus on how genomics and other emerging health technologies can provide more effective, personalised healthcare and deliver improvements in health for patients and citizens.

Sign up for the PHG Foundation’s newsletter: www.phgfoundation.org/signup
### Agenda

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<td>Registration and coffee</td>
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<td>09:45</td>
<td>Welcome and overview</td>
<td>Dr Hilary Burton&lt;br&gt;Director, PHG Foundation</td>
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<td><em>Plenary sessions</em></td>
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<td>10:00</td>
<td>Genomics in everyday clinical cardiology</td>
<td>Prof Perry Elliott&lt;br&gt;Professor of Cardiovascular Medicine UCL and St. Bartholomew’s Hospital, London</td>
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<td>10:30</td>
<td>Using genomics to improve reproductive health</td>
<td>Dr Tony Gordon&lt;br&gt;Lab Director, Genesis Genetics UK and VP, Cooper Surgical Global Genomics&lt;br&gt;Business Development</td>
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<td>11:20</td>
<td>Non-invasive genomics for cancer diagnosis and monitoring</td>
<td>Dr Nitzan Rosenfeld&lt;br&gt;Senior Group Leader, Cancer Research UK Cambridge Institute; Co-Founder and CSO, Inivata</td>
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<td>11:45</td>
<td>Q &amp; A</td>
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Parallel sessions
Please select one of the following parallel sessions

Session A - Genomics and infectious disease
This session will explore the ways in which genomics is poised to revolutionise the management of infectious diseases, through dramatic improvements in the effectiveness of outbreak investigation and the speed of disease diagnosis. It will also highlight opportunities to breakdown the barriers within our health system that are currently slowing the development and implementation of these technologies.

How do we catalyse the pathogen genomics revolution?
Dr Leila Luhesti
Head of Science, PHG Foundation

Translating microbial genomics into clinical practice
Dr Estée Török
Clinician Scientist Fellow, Department of Medicine, University of Cambridge

Applying rapid, portable nanopore metagenomics for improved diagnosis and management of infection
Dr Justin O’Grady
Senior Lecturer in Medical Microbiology, University of East Anglia

Session B - Genomics, health records and clinical care
This session will explore how novel approaches to genomic data sharing can be transformative in galvanising research and improving clinical care. It will also examine how the law, in attempting to protect individuals against harms associated with misuse of data, creates obstacles to these uses, and suggests how these issues may be addressed through building trust, improving data security, and developing ‘managed’ infrastructures and data-sharing processes.

Challenges to developing genomic data sharing for the NHS
Alison Hall
Head of Humanities, PHG Foundation

Linking genomics and health records
Prof David van Heel
Professor of Genetics, Blizard Institute, QMUL, and Chief Investigator and Joint Lead, East London Genes and Health

Patient benefits from integrating genomics and healthcare
Dr Mohammad Al-Ubaydli
CEO, Patients Know Best
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| 15:00  | **Panel session - The shape of things to come**<br>
*Perspectives and discussion on the role of science and society in shaping the future of personalised healthcare*<br>
Panel chair<br>
*Dr Philippa Brice*<br>
External Affairs Director, PHG Foundation

*Dr Anna Middleton*<br>
Head of Society and Ethics Research, Wellcome Genome Campus

*Dr Nick Lench*<br>
Chief Operating Officer, Congenica Ltd.

*Dr John Liddicoat*<br>
Philomathia Research Fellow, Faculty of Law, University of Cambridge

*Dr Ron Zimmern*<br>
Chairman, PHG Foundation

| 15:25  | Q & A                                                                |
| 15:50  | Summary and close                                                    |
| 16:00  | Reception<br><i>Delegates are invited to stay for drinks and informal networking</i> |
Dr Hilary Burton is the Director and one of the founder members of the PHG Foundation. She is Pro Vice President and Fellow of Hughes Hall, Cambridge. Hilary is a public health consultant with over 20 years experience leading work on the implementation of innovative technologies in UK health systems.

At the PHG Foundation, Hilary has been closely involved in the implementation of genomic technologies for screening programmes in the contexts of carrier screening, antenatal or newborn screening and has worked with major European Commission research programmes looking at the potential for susceptibility testing for common cancers and cardiovascular disease.
Plenary sessions

Genomics in everyday clinical cardiology

Prof Perry Elliott will overview the most common genetic forms of cardiac disease and discuss new clinical advances to diagnosis and genetic testing.

Perry is Research Lead of the Inherited Cardiovascular Disease Unit at the Bart’s Heart Centre, London, and Professor of Cardiovascular Medicine at UCL. He is cardiovascular lead for the North Thames NHS Genomic Medicine Centre, and a member of the Cardiovascular GeCIP (Cardiomyopathy subgroup). Perry has an international reputation in the field of heart muscle disease, authoring more than 300 peer-reviewed papers on the subject. He is a Fellow of the European Society of Cardiology (ESC) and chairs the ESC Guideline Task Force on Hypertrophic Cardiomyopathy and the Executive Committee for the European Outcomes Research Programme Registry on cardiomyopathies.

Using genomics to improve reproductive health

Genomics is revolutionising numerous aspects of reproductive health, from preconception genetic screening of couples, to preimplantation genetic screening (PGS) of IVF couples, to non-invasive prenatal testing and exome sequencing of the foetus. Dr Tony Gordon will discuss how genomics is currently used in reproductive healthcare and the future uses of genomics in this area.

Tony is the laboratory director for the Genesis Genetics UK labs and is leading the Cooper Surgical genomics companies global business development (outside the US). Tony is a PhD molecular cytogeneticist with over 20 years experience in molecular diagnostics, and is a UK registered Clinical Scientist. After working at the Institute of Cancer Research in the 90’s Tony moved to a number of companies in the diagnostics field, before joining Genesis Genetics.

Non-invasive genomics for cancer diagnosis and monitoring

Dr Nitzan Rosenfeld will explore the exciting opportunities that the extraction and analysis of circulating tumour DNA (ctDNA) offers cancer management, including tracking response to treatment, cancer progression and emergence of resistance to therapy.

Nitzan trained in Physics, and specialised in quantitative molecular biology, obtaining a PhD in the field of Systems Biology from the Weizmann Institute of Science. In 2005 he joined Rosetta Genomics, where he was head of Computational Biology and led development of molecular diagnostic tests. At the Cancer Research UK Cambridge Institute he leads a translational research group focusing on the application of ctDNA as a tool for cancer diagnostics and non-invasive genomics. In 2014, Nitzan co-founded Inivata, a clinical cancer genomics company that aims to harness the emerging potential of circulating DNA analysis to improve testing and treatment for oncologists and their patients.
How do we catalyse the pathogen genomics revolution?

The evidence that the widespread and systematic use of whole genome sequencing could significantly improve the management of infectious diseases in the UK and beyond is incontrovertible. However, the pace with which frontline clinical and public health services are adopting this technology remains slow. Dr Leila Luheshi will explore what we can do to accelerate progress towards patient and population benefit.

A former neurology researcher, Leila’s desire to see the results of biomedical research more rapidly and effectively converted into better health and healthcare for individuals led her away from the laboratory into her current role as Head of Science at the PHG Foundation. Leila specialises in writing accessible accounts of scientific advances and making persuasive cases for the policies needed to capitalise on them.

Translating microbial genomics into clinical practice

The ability to perform rapid, high-throughput whole genome sequencing using bench-top platforms represents a step-change in capabilities for diagnostic and public health microbiology laboratories. As the cost of sequencing continues to decline, the challenge will be to define when and where to apply this technology. Dr Estée Török will review its potential applications in the clinical microbiology laboratory and current barriers to implementation.

Estée qualified in Medicine from the Universities of Oxford and London, and did her specialist training in Infectious Diseases and Medical Microbiology at Oxford. In 2009 Estée was appointed as a Consultant in Infectious Diseases at Addenbrooke’s Hospital, Cambridge and set up two new clinical services – a bacteraemia consult service and an outpatient parenteral antibiotic therapy (OPAT) service. In 2011 Estée joined Professor Peacock’s research group at University of Cambridge as a Senior Research Associate and Honorary Consultant in Infectious Diseases and Medical Microbiology. She was awarded a Clinician Scientist Fellowship by the Academy of Medical Sciences and the Health Foundation in 2014.

Applying rapid, portable nanopore metagenomics for improved diagnosis and management of infection

Nanopore real-time sequencing technology has, for the first time, made it feasible to apply metagenomic sequencing to acute infection diagnosis. Dr Justin O’Grady will discuss how shotgun metagenomics sequencing has the potential to replace current methods, combining rapidity with comprehensiveness beyond that of culture or PCR.

Justin gained his PhD at the National University of Ireland, Galway (NUIG). This was followed by a post-doc at NUIG, two years in industry with Beckman Coulter and then a return to academia in 2010 as a Senior Research Associate at UCL. In 2013 he was appointed Lecturer in Medical Microbiology at UEA and was promoted to Senior Lecturer in 2016. His research focuses on the molecular diagnosis of pathogens and associated antimicrobial resistance in complex clinical syndromes such as sepsis, respiratory tract infections and urinary tract infections.
Challenges to developing genomic data sharing for the NHS

Implementing genomics into safe and effective care requires proportionate data sharing processes and frameworks. Alison Hall will address the challenges to genomic data sharing that are likely to arise from changes to UK, EU and international regulation, together with wider issues around data identifiability and public trust.

A qualified lawyer, Alison leads PHG Foundation’s regulation and ethics team. Recent work includes advocacy on data and device regulation that impacts innovations in healthcare. This includes appraising the impact of regulatory changes to EU data protection and medical devices regulation for clinical genetics services and medical research; working with stakeholders and statutory authorities to facilitate optimal data sharing policies in the NHS; and assessing how existing and future \textit{in vitro} diagnostic devices regulation might impact on IVD device and software development.

Linking genomics and health records

Prof David van Heel will discuss issues of trust, data security, scientific and data access, as well as genomics and e-health record linkage. He has particular excellence in this across East London with joined up data sharing of more than 1m NHS patients via the East London Genes & Health, a long-term DNA, e-health record and recall study of (British-) Bangladeshi- and Pakistani-origin residents.

David is Chief Investigator and Joint Lead for East London Genes & Health. He has been Professor of Genetics at Barts and The London School of Medicine and Dentistry, and Consultant Gastroenterologist at Barts Health NHS Trust since 2006. He also runs a Barts Health NHS Trust Gastroenterology Outpatient Clinic at the Royal London Hospital, as well as being Clinical Information Officer.

Patient benefits from integrating genomics and healthcare

The inspiration for the online Patients Know Best system was Dr Mohammad Al Ubaydli’s own experience living with a long-term illness and managing complex care across a range of health professionals. Mohammad will explain why patients are often the true experts on their own condition, and why putting control into their hands can benefit everyone.

Mohammad is founder and CEO of Patients Know Best. He trained as a physician at the University of Cambridge; worked as a staff scientist at the National Institutes of Health; and was a management consultant to US hospitals at The Advisory Board Company. He is an honorary senior research associate at UCL medical school for his research on patient-controlled medical records. In 2012 he was elected an Ashoka Fellow as a social entrepreneur for the contributions he has made to patient care.
Panel session
The shape of things to come

Panel chair
Dr Philippa Brice is External Affairs Director at the PHG Foundation where she oversees public and parliamentary affairs, strategic communications and partnering. Philippa originally trained in Natural Sciences (Pathology) at Christ’s College Cambridge before completing her doctorate at the MRC Laboratory for Molecular Biology and moving to work in the pharmaceutical industry. Her previous experience also includes science communication and health policy development.

Dr Philippa Brice
External Affairs Director, PHG Foundation

The Sanger Institute was founded on the principle that the human genome belongs to society. Since the completion of the Human Genome Project, the Sanger Institute has continued to develop a culture of data sharing and creating openly available resources, in order to maximise the impact of its science. Sarion Bowers will discuss how open science is changing scientific and innovation culture, and look at the challenges of sharing genomic data in a world of personalised medicine.

Sarion Bowers
Policy Lead, Wellcome Trust Sanger Institute

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Sarion is the Policy Lead for the Wellcome Trust Sanger Institute, a research organisation dedicated to using genomics and biodata to improve global health. She provides advice and guidance to researchers, legislators and policy makers on bioethics, research regulation and science policy. Originally a research scientist, Sarion now specialises in science and technology policy relating to genomics. Sarion has a particular interest in the ethical, legal and societal issues that arise from the use of genomics in the clinic and the development of new genomic technologies, including genome editing.

Dr Nick Lench has an interest in moving genetics and genomics into mainstream medicine. He will discuss some of the challenges facing the NHS in effecting this transition as well as the requirement to link and share clinical records and outcome data.

Dr Nick Lench
Chief Operating Officer, Congenica Ltd

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Nick is Chief Operating Officer at Congenica, a clinical genomics company pioneering genome based medicine. He is also Chair of the Joint Committee on Genomics in Medicine, Honorary Reader at the UCL Institute of Child Health and has over 25 years experience in human molecular genetics. Nick was previously Director of the NE Thames Regional Genetics Service at Great Ormond Street Hospital for Children, with responsibility for the strategic and operational management of a genetics service that provides DNA diagnostic testing to a population of approximately 4.5m people. He was awarded a personal chair in Medical Genetics at Cardiff University in 2005, and was founding CEO of London Genetics Ltd and Programme Director at Oxagen Ltd.
Global patent protection for new diagnostic tests is currently in a state of uncertainty, which has been primarily brought about by a recent series of US Supreme Court decisions. Dr John Liddicoat will discuss the details of these US decisions, how research is needed to shed light on how these cases are affecting diagnostic development, and how smart intellectual property policy will play a key part in the realisation of personalised healthcare.

John is interested in the development and use of new technology because it has the ability to drive economies forward, increase public welfare and solve important social problems. His research focuses on the ability of patent law to meet its welfare-enhancing goal of accelerating the creation of new technology. John is currently working on a research project titled, “Realising Genomic Medicine: Intellectual Property Issues Beyond the ‘Old’ DNA Patent Debates”.

Dr Ron Zimmern will outline the consensus view from a recent international summit of population health experts on four essential ‘disruptive leaps’ needed for personalised healthcare to keep pace with scientific advances and engage with citizens. He will also ask the question with whom does the responsibility for the delivery of personalised healthcare really lie.

Ron is the founder of the PHG Foundation and Chairman of the Board of Trustees. Ron has enjoyed a distinguished career in medicine, public health and policy. He has been Director of Public Health for Cambridge and Huntingdon Health Authority and pioneered public health genomics internationally. Ron has a degree in law and an enduring interest in medical law and ethics; he is a Fellow of Hughes Hall, and holds an honorary Professorship at the University of Hong Kong. National roles in genomics, commissioning and screening have included membership of the Genetics Commissioning Advisory Group; National Genetic Testing Network Steering Group, Joint Committee of Medical Genetics of the Royal Colleges; Council for the British Society of Human Genetics; and Chairman of the HTA Diagnostic and Screening Panel.