The future of personalised healthcare should be focused on helping individuals, health professionals and policy-makers work together. Harnessing technology should help, but the whole endeavour must also be grounded in sound science and practical reality.

Philippa Brice, Oct 2016

Annual review 2016

PHG Foundation is a non-profit think tank, working to achieve the fast, effective and responsible application of biomedical and digital health technologies to bring benefits to all patients and citizens.

In 2016 we...

- published 10 papers in academic journals
- produced 6 reports
- submitted evidence to 6 consultations

covering issues related to the clinical implementation, regulation and ethics of topics including:

- data security
- accelerated access to medicines
- genome editing
- non-invasive prenatal testing

...speaking up for better healthcare in more than 60 opinion pieces and throughout all our activities

Is it acceptable that access to the advances in safety and effectiveness offered by genomic medicine is accelerated only for those with deep enough pockets to afford it?

Leila Luheshi, Nov 2016

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We have been working with Genomics England on their ‘Big Conversation’ engagement programme on the potential of genomics and issues likely to arise in its implementation within health services.

We brought leaders from NHS England and Genomics England together with senior clinicians from a range of medical specialties to discuss the implementation of genomics across the NHS. We followed ‘conversations with clinicians’ with a second conversation at the House of Lords, where more than fifty parliamentarians and expert stakeholders, discussed the purpose and progress of the 100,000 Genomes Project and the implications for patients and the NHS.

Our summary of the conversations informed the final report from Genomics England on all the ‘Big Conversation’ meetings.

On behalf of our Chairman, Dr Ron Zimmern, we coordinated a summit of international population health experts to address the systemic challenges of achieving the disruptive agenda of precision healthcare. In a communique, Paths to precision health: act now, the convening group calls for active leadership to harness new technologies to drive the public’s participation in its health.

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It is unlikely that microbiome analysis will ever be useful in isolation as a tool to improving health. Instead, if it is used as part of an integrated approach to personalising healthcare that leverages knowledge about a wider range of biomarkers, it may in time prove useful.

Sowmiya Moorthie, April 2016

Healthcare futures

Our healthcare futures research includes succinct overviews, presented in blogs and infographics, of areas as diverse as blockchain and machine learning, synthetic biology and the microbiome. They set out for non-experts which technologies are really ‘hot’ and which (once we’ve cut through the hype) are not.
As the secretariat to the All Party Parliamentary Group on Personalised Medicine, PHG Foundation is cementing relationships with parliamentarians excited by the potential science and technology have for health and personalised medicine.

**Making the most of personalised medicine**

Opened by then Life Sciences Minister George Freeman MP, the inaugural event of the APPG on Personalised Medicine, attracted over 100 parliamentarians and stakeholders. The APPG on Personalised Medicine aims to enable swift patient benefit from scientific and technological advances, including in relation to common diseases. The group also expects to work with other APPGs on issues of common interest related to science and health policy.

**In 2016 we have been strengthening our ties with**

- Cambridge University Health Partners
- Cambridge Institute of Public Health
- Genomics England
- Hughes Hall, University of Cambridge
- Chinese University of Hong Kong
- Faculty of Law, University of Cambridge
- Association for Clinical Genetic Science
- Microbiology Society
As technology advances, increased data access and connectivity presents multiple opportunities, but also considerable challenges. The UK - and European - policy arena has been highly exercised by data security and usage.

Providing the evidence

Data sharing for patient benefit

The complex debates around the use of personal health data and the need to develop proportionate and feasible data sharing policy fuelled a busy year of advocacy for both our science and humanities teams including speaking engagements at the Human Variome Project biennial meeting, the European Society of Human Genetics annual conference, and a presentation to the Genomics England Ethics Advisory Committee.

Having commended the recommendations of the PHG Foundation and ACGS* joint report, Data sharing to support UK clinical genetics and genomics services, in the latest Caldicott Review on data security, consent and opt-outs, the National Data Guardian's Office subsequently invited PHG Foundation to collaborate in planning and delivering their first evidence session, focused on genomic data sharing for clinical care.

...a recent joint report from the PHG Foundation and the Association for Clinical Genetic Science makes a number of commendable recommendations.

Dame Fiona Caldicott, National Data Guardian for Health and Care Review of Data Security, consent and Opt-outs

*Association for Clinical Genetic Science
Our in-house expertise in relevant regulatory systems combined with a strong track record of collaboration on EU funded programmes placed us in pole position to win new funding as part of the EPIC-CVD study into novel biomarkers for cardiovascular disease improving risk prediction.

Genomics and heart disease

As part of the EU-funded EPIC-CVD programme we investigated the regulatory implications of using novel risk stratification tools incorporating genomic information to deliver increasingly sophisticated data on personal disease risk. In our report to the EPIC-CVD group, Genomics and risk stratification for cardiovascular disease: regulatory implications, we examine the complex regulatory systems that exists in different countries – taking France, Germany, the Netherlands and the UK as examples - and evaluate the impact of current and forthcoming European legislation.

See also:

Whole genome sequencing for breast cancer risk testing, our 2016 report on the impact of routine testing for heritable conditions using whole genome sequencing.
Our work on the implementation of pathogen genomic technology to aid surveillance and control of infectious disease outbreaks, published in 2015 in *Pathogen Genomics Into Practice*, continues to generate considerable interest.

**Big Data or bust**

Since issuing our roadmap for implementation we have been working closely with the Microbiology Society on data sharing to aid surveillance and curtail outbreaks, a fruitful collaboration marked with a lively lunchtime panel session - *Big Data or bust* - run by PHG Foundation at the society's annual conference in March.

**We have the technology – but can we afford it?**

This is the question addressed by PHG Foundation when we partnered with University College London and the University of Oxford to evaluate the cost impact of using whole genome sequencing (WGS) for the detection and isolation of mycobacteria spp. in the context of tuberculosis in humans. The results, which provide further evidence on the financial viability of WGS-based diagnostics, will inform the Health Innovation Challenge Fund (HIFC) recommendations on implementing WGS to tackle infectious disease.
In navigating the maze of data protection law and standards, our regulation and ethics team uncovered significant flaws in the EU data protection regime as it pertains to genomic data, which raise urgent questions for policy makers. Instead of developing ever-more sophisticated methods of anonymisation in response to increasing threats of re-identification there is a need for policy makers to have an overview of the issues raised by using genomic data and some possible solutions.


**The truth behind ‘anonymisation’**

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**See also:**

- Our series of blogs on data regulation and ethics by Alison Hall
- Our consultation response on the National Data Guardian (NDG) for Health and Care’s review of data security, consent and opt-outs

The urgent challenge is for all those who want to see a health service with high quality, safe, effective and evidence-based care to be much more vocal and resolute in demonstrating that data sharing is a vital and necessary part of keeping well and an integral component of healthcare.

Alison Hall, July 2016
We ended 2016 on a high note, with a full day of exploration and discussion of some of the most promising technologies emerging into healthcare at our Life Sciences & Society conference on personalised healthcare.

Close to one hundred industry representatives, lawyers, researchers, clinicians and policymakers gathered at our sold out conference to discuss the potential of genomics to deliver more personalised healthcare – and the implications for policy, patients and society.

In the morning experts from cardiology, cancer and reproductive health enlightened delegates on the exciting advances genomics is bringing to diagnoses and treatments in their respective fields, while the final session included lively discussions on the conflict between access to data and incentivisation, and building trust in order to enable transparency.

The programme for the Life Sciences & Society 2017 will be on the website in early January.

“Very glad that I attended. Talks were pitched at a variety of levels to enable a diverse audience to engage and stay involved and enthused. Thank you for recognising the importance of balancing clinical, scientific, socio-cultural and policy perspectives.”

“Breakout session on genomics, health records and clinical care was excellent.”
With a special focus on science- and technology-propelled innovations relevant to health systems we have been providing clients with a range of consultancy services tailored to meet their organisational needs.

Our consultancy services

Initially engaged by a commercial client on a one-off basis to provide insight into the implementation pathways for their technology, we were pleased to extend the contract to offer bespoke weekly briefings on relevant regulatory and policy developments.

Our productive relationship with the UKGTN continued with a commission from them to update their original UKGTN guide to centres with specialist services for rare genetic disorders, which was published in Jan 2016. They have since commissioned us to design and publish a third edition.

NHS National Services Scotland (NHSS)* commissioned PHG Foundation to undertake a literature review in the field of genetics and molecular pathology services to inform the NHSS review of future provision of these services to meet the needs of the country’s population.

*The report produced by PHG has been highly informative to NSD, as commissioners of the service. It has also provided the Independent Expert Review Group with a solid evidence base to make recommendations*

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