making science
work for health

PHG FOUNDATION

ANNUAL REVIEW 2023

Message from the Director

I have long been an admirer of the PHG Foundation's work, so it is a great privilege to be entrusted with the role of Director. Between stubborn threats to population health and the rapid pace of research and innovation, PHG's mission, 'to make science work for health' is, if anything, more important now than it has been at any point in our history.

Since taking over from Mark Kroese in June, I have enjoyed fantastic support from my team of experienced and energetic colleagues. It has been a phenomenally busy period, even by PHG's standards, which has resulted in some important and influential analyses of emerging health technologies, from polygenic scores to synthetic data, and the complex policy challenges they raise.

Earlier in the year we marked our 25th anniversary with a policy summit and gala dinner. It was a great opportunity to take stock of the emergence of genomics and related technologies, and the rules, policies, practices and institutions that we have placed around them to harness them for public benefit.
We are looking ahead to the future with refreshed determination to make science work for health, for all.

Dr Peter Mills
Director

“Making science work for health will always rely on getting the right institutions, policies and practices in place. This is where PHG is uniquely placed to contribute – and we must ensure that we are able to respond to the evolving environment.”

- Pete Mills

Reports

Project DELTA

The innovative Project DELTA (integrateD diagnostic solution for EarLy deTection of oesophageal cAncer), came to a close at the end of the year. Throughout the three-year duration of the project, we have been leading ethical and legal research into personalised risk prediction and AI driven pathology to detect oesophageal cancer, and its precursor, Barrett's oesophagus.

Conclusions from our research are set out in the two reports we delivered for our partners on the DELTA steering group.

Workshop report on adopting a risk tool for the stratification and predictive prevention of oesophageal cancer

Published March 2023

Human involvement in AI-driven digital pathology pathways: ethical and legal considerations

Published December 2023

Both reports are available on our website, as is a DELTA position statement in which the partners offer a set of recommendations to policymakers.

Are synthetic health data ‘personal data’?

We were commissioned by the Clinical Practice Research Datalink team at the MHRA to address the emerging question of the status of synthetic health data under data protection law. Our report is the first of its kind to consider this topic and highlights ways that synthetic data developers, researchers, regulators and policymakers should respond to current ambiguity and move forwards with appropriate regulation for this important area.

Published July 2023

Heat, health and human genetics

In our independent report, we discuss current understanding of the direct threats of heat to health, and potential ways in which genetic information on heat responses could be applied to manage the future health burden associated with heat and aggravated by climate change.

Published December 2023

Polygenic scores

Evaluation of polygenic score applications

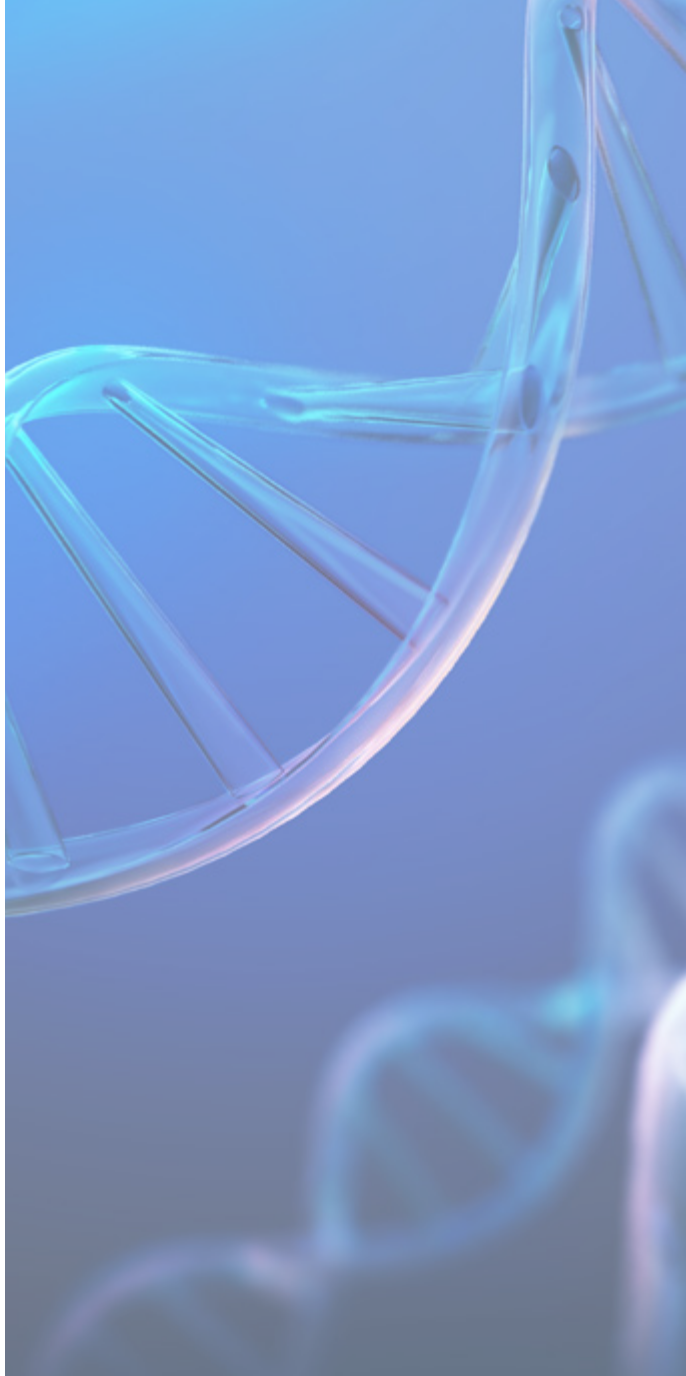
In our most recent report on polygenic scores we analyse the application of the principles of medical test evaluation to PGS-based products. By bringing together concepts from different fields and using the ACCE framework we show there currently is a lack of robust evidence for the utility and validity of polygenic scores as independent tests.

We recognise that polygenic scores are likely to be useful under certain circumstances, but identifying these and creating optimal systems for their use requires a focused approach to evidence generation and appraisal which is currently lacking.

Published June 2023

As debate around polygenic scores continues we homed in on the fundamentals in a report, podcast episode and in a suite of briefings that covered:

- Unpacking polygenic scores
- Application of polygenic scores in healthcare
- Polygenic score analysis: the test pipeline
- Regulating polygenic score devices and tests
- The path to using polygenic scores in healthcare



Working with partners

We have worked with a number of external clients and partners this year. We've delivered projects for the Medicines and Healthcare products Regulatory Agency (MHRA), Genomics England, InSilicoUK, and UK Research and Innovation (UKRI). As well as an important source of funding for us, such projects present opportunities to extend our networks and keep up to date with the latest thinking and activity on health technologies as they emerge from research and before they affect patients.

Medicines & Healthcare products Regulatory Agency

Royal Academy of Engineering

INSILICO UK

PROPHET
a PeRsOnalized Prevention roadmap for the future HEalThcare

CMEL
The University of Hong Kong Centre for Medical Ethics and Law

Genomics england

EXACT

WYNG FOUNDATION

SQW

Commissioned and collaborative work completed this year includes:

The regulation and governance of lifetime genomic data

Funded by Genomics England, we examined the ethical and legal considerations that stem from the Newborn Genomes Programme.

Data to early diagnosis and precision medicine challenge

A five-year collaboration with public policy consultants SQW, commissioned by UKRI, came to an end with the delivery of the final report in which we contributed to the evaluation of two strands of the funded challenge: genomics and the HDR UK health data programme.

Journeys, experiences and best practices on computer modelled and simulated regulatory evidence

We worked with InSilicoUK, the MHRA and the Royal Academy of Engineering on a workshop and report identifying common themes and next steps for the adoption of computational modeling and simulation in regulatory pathways.

Report published December 2023

Our global perspective

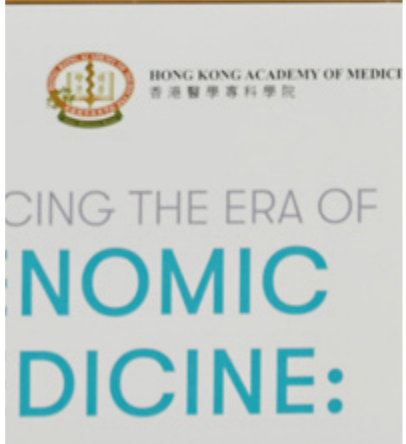
Our mission to make science work for health extends to health systems beyond the UK. Often our work has application to local health contexts in multiple countries (for example our series of reports for FIND last year). This year, however, we had the honour of collaborating with the University of Hong Kong on the evaluation of the Hong Kong Genome Project, as well as continuing our collaboration with the Hong Kong-based Center for Medical Ethics and Law.

We met a key milestone this year working with colleagues from the Centro de Investigación Biomédica en Red (Spain) as part of the European PeRsOnalized Prevention roadmap for the future of HEalThcare (PROPHET) project, with the delivery of a report on biomarkers for risk prediction and stratification for cancer, cardiovascular and neurodegenerative diseases. These results were well received and will shape our research, and that of our partners going forward in this project.

Although no longer part of the European Union, changes in EU law and regulations still have implications for UK researchers, as we discussed in our work examining the development of European health data spaces.


Hong Kong Genome Project
香港基因組計劃
請在此等候


The European
Health Data Space


HONG KONG ACADEMY OF MEDICI
香港醫學專科學院
CING THE ERA OF
NOMIC
DICINE:

Making science work for health

The **Making science work for health** policy summit was to be the highlight of spring 2023, rather than autumn 2022 as orginally planned. Professionals from research, healthcare, government and industry gathered to hear presentations from leaders from across the health and policy landscape.

Speakers

- Prof Dennis Lo. Director, Li Ka Shing Institute of Health Sciences
- Dame June Raine, Chief Executive, Medicines and Healthcare products Regulatory Agency
- Prof Dame Sue Hill, Chief Scientific Officer, NHS England
- Prof Gil McVean, Founder and CSO, Genomics plc
- Prof Clare Turnbull, Professor of Translational Cancer Genetics, Institute of Cancer Research
- Dr Richard Scott, Chief Medical Officer, Genomics England
- Dr Sarah Byron, Associate Director of the NICE Diagnostics Assessment Programme
- Prof Sally Sheard, Head of the Department of Public Health and Policy, University of Liverpool
- Prof Sir John Burn, Professor of Clinical Genetics, Newcastle University

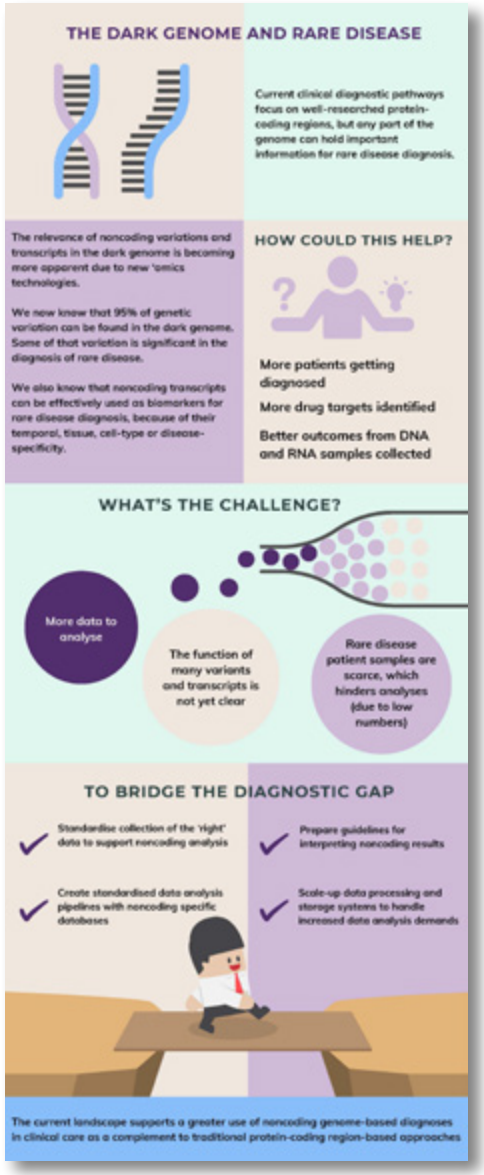
Celebrating 25 years

Long-time friends and supporters gathered together at Trinity College, Cambridge to celebrate 25 years of the PHG Foundation.

Members of The Sixteen opened the gala dinner with a stunning rendition of *Laudi Spirituali*.

Prof Dame Sally Davies, Master of Trinity College, gave a warm welcome to guests from the UK and around the world including Canada, Hong Kong, Italy and France. Luminaries of health policy, academia, law and medicine joined speakers from the policy summit which was held earlier that day.

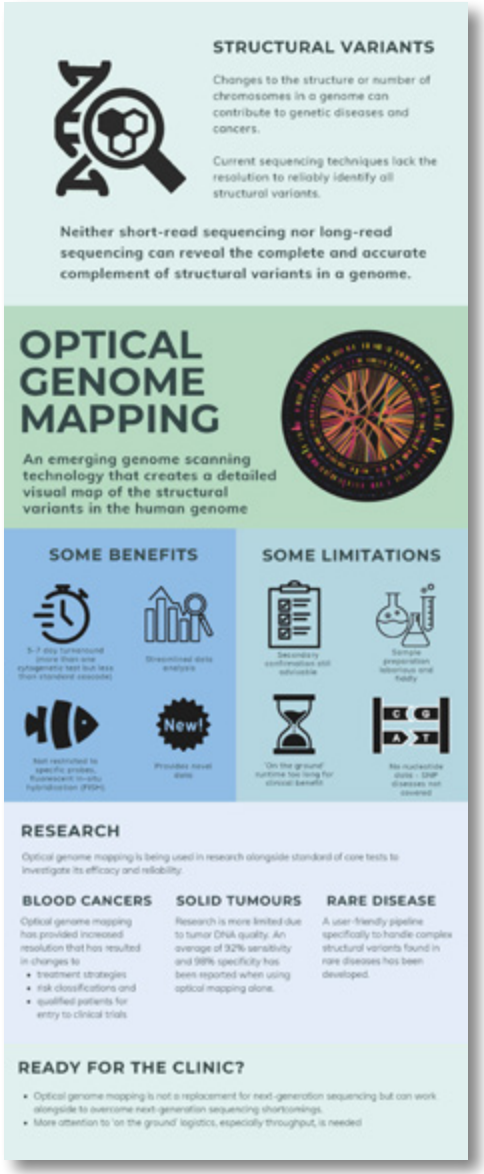
New science explained



We continue to monitor novel technologies emerging from the laboratory, sharing our evaluations of their readiness for use in routine patient care in briefings, infographics and the **Making science work for health** podcast.

For those newer to genomics in healthcare, or for professionals needing a concise overview on the state of 'omics and human health science, we are relaunching our innovative health infographics and have added three new podcast episodes.

With these higher-level explainers, we aim to engage more people in the conversation about how we use technology to prevent and treat common diseases.



New podcast episodes:

- Explaining polygenic scores - and their limitations
- What is xenotransplantation?
- What is long-read sequencing?

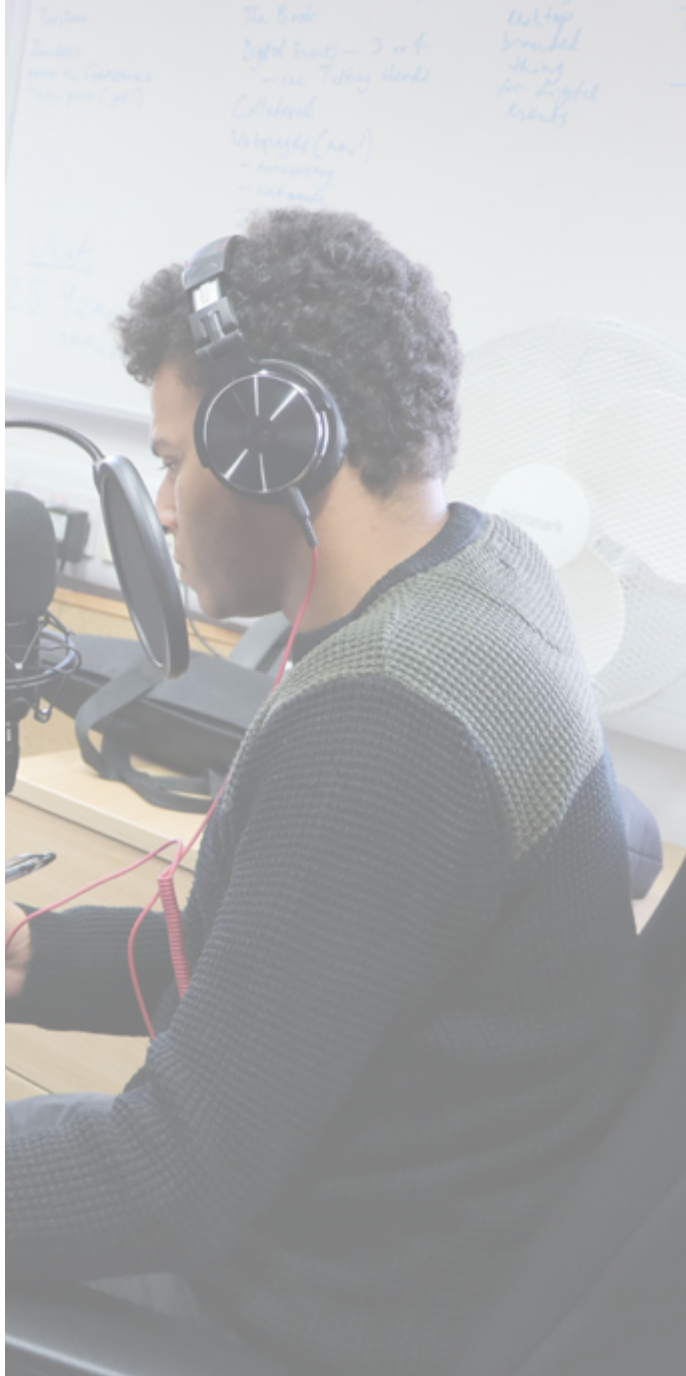
All our digital products are freely and publicly available via our website.

Briefings

2023 was a bumper year for PHG briefings, boosted by the set of five briefings explaining the current level of evidence for polygenic scores, and what policymakers, researchers and regulators need to do to address the gaps.

We issued an essential briefing on the implications of the European Health Data Space for UK research, fulfilling a need for information about this important European initiative.

By the end of the year, three new briefings, commissioned by the WYNG Foundation, and complemented by new infographics, were ready for release.



External engagement

Key

- Journal publication
- Speaker or panelist

January - March

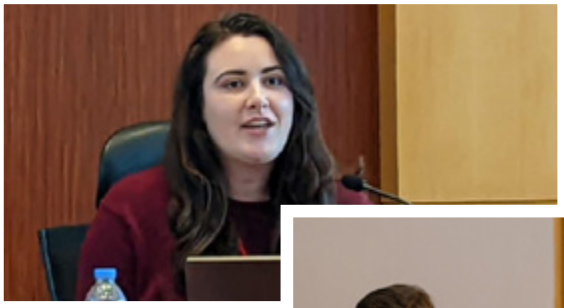
- Sowmiya is at the **Festival of Genomics,** speaking on polygenic scores
- *Lessons from the pandemic for the future regulation of confidential patient information for research* - Journal of the Royal Society of Medicine
- Laura is invited to speak on current PHG Foundation activities at the **Clinical Geneticists Seminar** in London
- *Public Preferences for Determining Eligibility for Screening in Risk-Stratified Cancer Screening Programs* - Society for Medical Decision Making
- *Proactive breast cancer risk assessment in primary care* - British Journal of Cancer
- *Ethical and legal considerations influencing human involvement in the implementation of artificial intelligence in a clinical pathway* - Frontiers in Digital Health

April - June

- Mark speaks on the Hong Kong Genome Project at the **Hong Kong Academy of Medicine Symposium**
- Colin gives **three** presentations at the AI conference organised by **Centre for Medical Ethics and Law of the University of Hong Kong**
- Laura speaks on influencing policy to the **UK Cancer Genomics Group**
- Colin speaks on personalised prevention and returning genetic test results with familial consequences - **KU Leuven, Belgium**
- **European Society of Human Genetics** - Chantal joins as workshop co-ordinator and Colin co-chairs a session on data altruism and future of data sharing in genomics
- Alison speaks on ethical issues in prenatal genetic testing at **Royal College of Obstetricians and Gynaecologists World Congress**
- Alison is a panelist at the **RSM's Ethics and regulation for AI in health**

July - September

- Sowmiya presents on polygenic scores to the EU funded **INTERVENE** project
- *Are we nearly there yet? Starts and stops on the road to use of polygenic scores* - Journal of Community Genetics



October - December

- *Ethical and legal implications of implementing risk algorithms for early detection and screening for oesophageal cancer, now and in the future* - PLOS ONE
- *A Principle-Based Approach to Visual Identification Systems for Hospitalized People with Dementia* - Bioethical Inquiry
- Pete gives a keynote talk at the **Cambridge Public Health Showcase**
- Pete speaks about our current activities at **the Healthy ageing and precision medicine conference**, Abu Dhabi
- Colin, Pete and Tanya each give presentations at the **Centre for Medical Ethics and Law of the University of Hong Kong** annual conference
- Sowmiya speaks on polygenic scores at the **Future of Predictive Genomics**, Denmark
- Elizabeth presents key findings from the synthetic data report to **Pan-UK Steering Group**

People

Tanya Brigden

Dr Mark Kroese

Dr Laura Blackburn

Dr Chantal Babb de Villiers

Heather Turner

Dr Sowmiya Moorthie

Dr Colin Mitchell

Alison Hall

Dr Hayley Wilson

Dr Elizabeth Redrup Hill

Rebecca Bazeley

Bhavya Krishnan

Dr Chaitanya Erday

Ofori Canacoo

Nicola Sparks

Sheila Rush

Governance

As a linked exempt charity of the University of Cambridge, we are proud to have a distinguished group of trustees providing support and strategic oversight to help us deliver our mission:

- **Prof Patrick Chinnery -** Head of the Department of Clinical Neuroscience, University of Cambridge (until October 2023)
- **Prof Nita Forouhi** - Professor of Population Health and Nutrition, University of Cambridge
- **Dr Anthony Freeling** - Acting Vice Chancellor, University of Cambridge and former President of Hughes Hall
- **Mr Andrew Hutton** - Director, AJ Hutton Ltd
- **Prof Mike Inouye** - Professor of Systems Genomics & Population Health, University of Cambridge (from October 2023)
- **Prof Patrick Maxwell** - Regius Professor of Physic, University of Cambridge
- **Prof Liba Taub** - Professor Emerita of History and Philosophy of Science, University of Cambridge and Director of Research at the Whipple Museum of the History of Science
- **Dr Ron Zimmern** - Founder and Chairman of the PHG Foundation

Sustainability

The health of current and future populations is affected by today's actions. As a responsible member of the UK public health community, PHG Foundation works to make a difference by understanding the facts about climate change and taking a responsible approach in our projects, wherever possible and appropriate, to help health systems deliver 'greener' models of care; and by setting an example by employing carbon reduction measures in our own day-to-day activities in the workplace

Equality, diversity and inclusion

Equality of opportunity, valuing diversity and compliance with the law benefits both individuals and organisations. At the PHG Foundation, we recognise people as individuals with diverse opinions, cultures, lifestyles and circumstances and strive to ensure that all our employees are valued and treated with dignity and respect. We want to encourage everyone in our business to reach their potential and seek to develop the skills and abilities of all our employees.

Finances

The PHG Foundation is funded by philanthropic donations, primarily from the Hatton Trust and the WYNG Foundation. We are not funded by the University of Cambridge. Other income comes from grants, collaborations, and commercial and public sector consultancy. We also have a modest investment portfolio. Most of our spending is on charitable activities, which includes our work programme and staff costs.

2022-2023 income: £1,259k

2022-2023 outgoings: £1,124k

For new PHG Foundation insights and events:

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- Search PHG Foundation on Spotify, Buzzsprout etc.
- Sign up for the newsletter www.phgfoundation.org