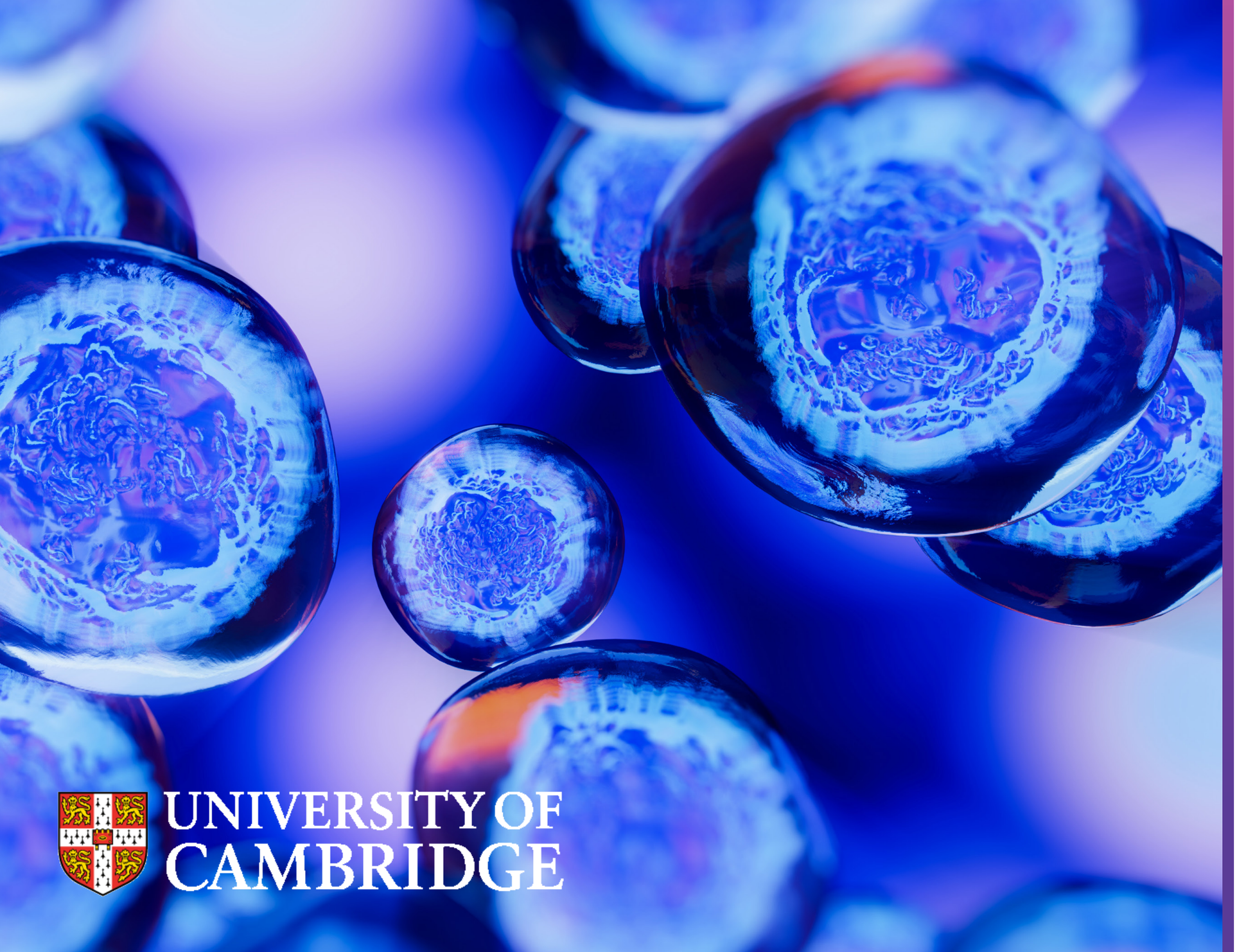


UNIVERSITY OF CAMBRIDGE

PHG FOUNDATION

making science work for health

2021

The PHG Foundation is a charity that provides multidisciplinary analysis of biomedical innovations and ideas to inform healthcare policy and practice, helping everyone who wants to see good science make healthcare better, faster

PHG Foundation
2 Worts Causeway
Cambridge
CB1 8RN
UK

Tel: +44 (0)1223 761 900

Director's message

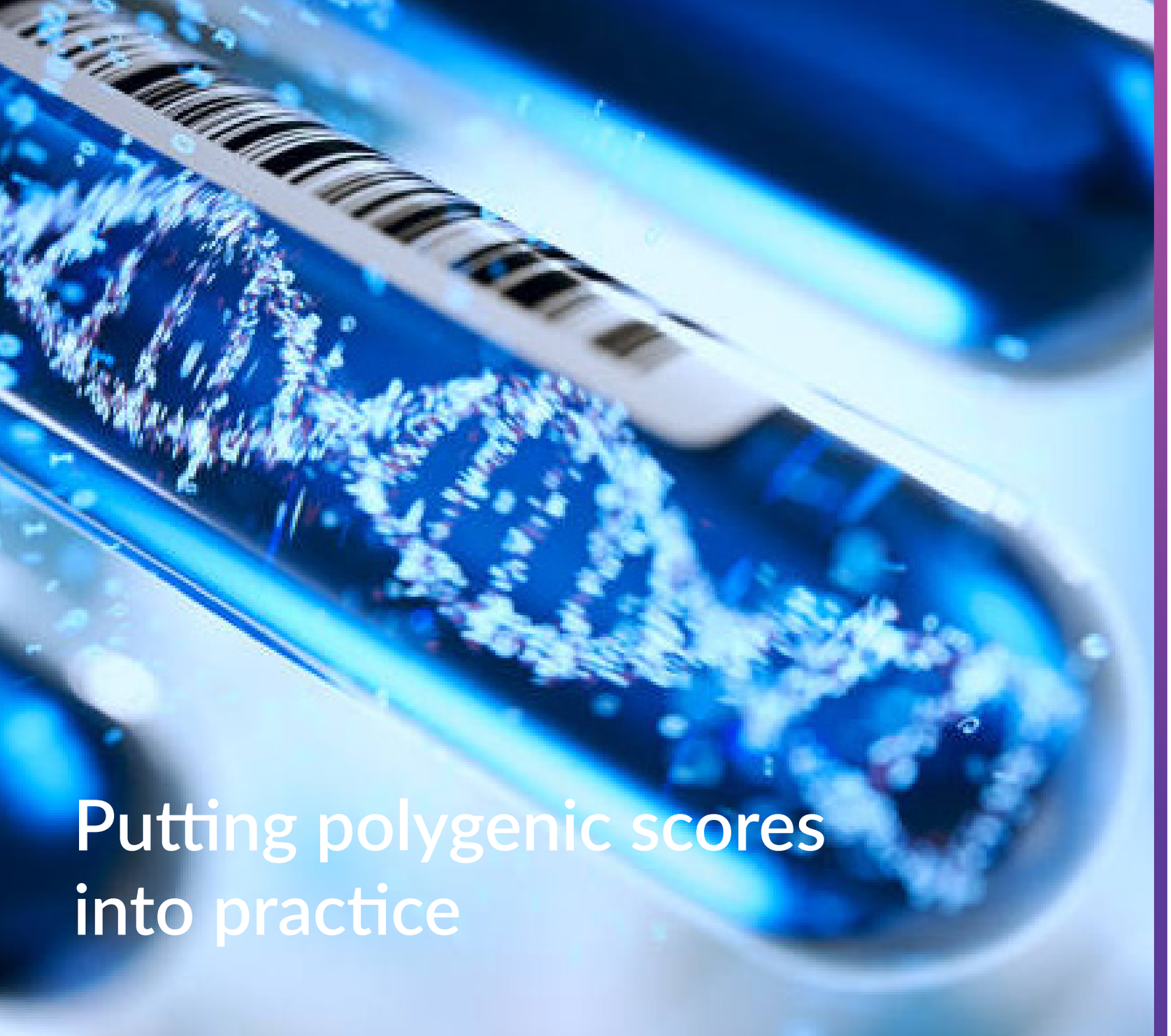
Over the last year, the imperative to harness science and technology and provide practical solutions to real-world health issues has been further underlined. We have seen some remarkable developments in diagnostics, vaccines and treatments; in digital transformation; and in the use of new forms of non-invasive and near-patient testing and remote monitoring.

Throughout 2021, the PHG Foundation team worked on a range of policy issues, from the prospects for new areas of science to benefit health, and new applications of current technologies, to the ethical and legal implications of biomedical developments. Besides our own horizon-scanning, research and analysis, we added perspective and value to the efforts of several important national and global partner organisations.

The Foundation is supported by a distinguished Board of Trustees, who provide strategic oversight and guidance. In 2021 we said goodbye to Ian Peacock, who stood down as a Board member after more than ten years; he was an unfailing source of invaluable advice and scrutiny, and will be greatly missed. We were however delighted to welcome Andrew Hutton as a new trustee. He is currently a member of the Governing Body of the Lister Institute of Preventive Medicine, a Director of the Brunner Investment Trust and has extensive experience in the financial sector.

As we enter 2022, our own 25th anniversary year, it is clear that our ongoing mission to support policy makers in making science work for health remains as important as ever. With pressing health system and public health challenges ahead, we will continue to provide the ideas, insights and intelligence that will help put innovative solutions into practice.

Dr Mark Kroese
Director



Putting polygenic scores into practice

The risk of developing most diseases is polygenic - influenced by multiple genetic variants, as well as other environmental and individual factors. There is considerable debate as to how far the use of polygenic scores could help to improve risk prediction and prevention for conditions such as cancer, diabetes or heart disease. Our work programme in this area seeks to understand the most important implications of new research for clinical and public health practice and policy.

In 2021, our first report on this area examined how useful polygenic score analysis could really be in routine healthcare, particularly what evidence is needed to demonstrate clinical utility. This is an essential first step towards use in health services.

Even where the scientific links between polygenic scores and disease risk are clearly demonstrated, clinical utility depends on a range of further factors, including how, where and when the scores are used. We found that structuring research into polygenic scores to deliver the right evidence of utility could speed up progress towards patient benefit.

Our second report looked ahead to consider what could happen once polygenic scores have proven clinical utility for prevention of a specific type of disease. Building on interviews from a range of experts, we demonstrated how polygenic score analysis could be incorporated into the current NHS Health Checks programme to reduce the impact of cardiovascular disease.

Our analysis reveals the potential implications for patients, health professionals and policy, highlighting the actions needed to ensure that more personalised risk prediction offers benefits without causing unintentional harm.

New horizons for healthcare

The PHG Foundation is widely recognised for our expertise in the different uses of genomics for health, developed over the last twenty five years. However, there are many other new and emerging areas of science and innovative technologies that have the potential to change the way in which we manage our health and treat disease. Some may be used in combination with genomics and other 'omics approaches, or otherwise contribute to data-driven healthcare; some may be relatively independent.

Keeping ahead of biomedical developments allows us to identify the emerging policy issues that need to be addressed - typically well in advance - to ensure effective uptake of useful new technologies within health services, and maximise the benefits for health professionals, patients and the wider public.

Our *Innovations for Health* programme of horizon-scanning research and analysis looks at innovations in detail, identifying those with the greatest potential to improve health and assessing the likely timescale for their impact. We consider the practical, policy and wider social issues that innovations may pose; for example, possible disruption to current care pathways and systems; any ethical dilemmas associated with use; and regulatory implications.

This programme builds on our extensive network of professional experts in different fields, and regular review of the scientific, medical and policy literature. Our findings not only inform and direct our wider work, but are also shared with wider audiences via explainers and other knowledge products. In 2021, our most popular publication was our policy briefing on RNA vaccines, which was viewed over a million times.

Digital twins - the future for medicine?

Conceived in the field of engineering, **digital twins** are an approach to modelling now being explored as a new approach to personalised medicine. Virtual avatars created to mirror data from an individual could be used to predict and model health outcomes, and plan treatments and interventions. However, our analysis of current evidence suggests that it has very limited utility for biomedical research or practice at the moment.

Control of confidential patient information

In the past, individual consent or approval from the Health Research Authority was needed before confidential patient information could be made available to researchers. When the pandemic hit in 2020, special Control Of Patient Information (COPI) notices were issued to enable rapid access to high quality data for crucial COVID-19 research. These COPI notices remove the normal legal approval requirements, speeding up the release of data for research to help understand how the virus spreads and causes illness, and to develop new tests and treatments.

We were commissioned to undertake independent research into the impact of COPI notices on genomics and related medical research, funded by the Department of Health and Social Care Policy Research Programme. Our research showed that the new streamlined approaches to patient data sharing had very positive effects for public interest research - so should there be permanent reform to how such information is shared?

Our ethical and legal analyses and review of public-attitudes suggest that whilst streamlined processes have already catalysed some improvements beyond the COPI notices themselves, there are significant considerations for policymakers in making further reforms. Most importantly, developing and demonstrating the trustworthiness of those involved in data sharing, through increased transparency and meaningful engagement, to ensure public and professional confidence in use of data for research.

FUNDED BY
NIHR | National Institute for Health Research

A centre of excellence

A unique policy think tank, with a sole focus on making science work for health, we are proud to be a linked exempt charity of the University of Cambridge. Whilst we operate internationally, the PHG Foundation also contributes multidisciplinary expertise to initiatives closer to home.

The Da VINCI project led by THIS Institute is developing a visual identification system for people with cognitive impairment in institutional settings. As our contribution to this important work for 2021, we analysed the ethical and legal issues posed by the use of visual aids to identify people in hospitals who have a diagnosis of dementia, and developed a set of principles to help guide the development and implementation of such systems in the next phase of the project.

Our experience in appraising the impact of new technologies is contributing to project DELTA, a multi-centre collaboration between the Universities of Cambridge and Oxford, Kings College London, and the diagnostics company Cyted. This ongoing initiative aims to improve personalised prevention and survival for oesophageal cancer through the use of innovative technology and artificial intelligence. We have been exploring potential ethical, legal and regulatory challenges, starting with a series of stakeholder workshops to consider the implications of this new approach.

Working with

THIS.Institute The Healthcare Improvement Studies Institute

Continuing the battle against COVID-19

Over 2021, we undertook research commissioned by FIND, the global alliance for diagnostics. This work looked at how the use of genome sequencing technologies for global surveillance has facilitated identification and characterisation of the SARS-CoV-2 virus, tracking the emergence of viral variants and informing public health actions to mitigate the impact of the COVID-19 pandemic in different countries.

We found that these methods are complex and developing rapidly; a lack of standardisation in how they are used further complicates the global scientific landscape for how and where they are used in pathogen control. Our report for FIND also explored the importance of global data sharing, and how certain countries could support others in ongoing efforts against the pandemic.

A second report looked specifically at the impact of new viral 'variants of concern' on diagnostics, vaccines and other public health tools. Variants of concern are viral strains that have potentially dangerous genetic changes, for example by making them more transmissible (such as the Omicron variant), or likely to cause more serious illness. Our work set out how genome sequencing can support continued monitoring, evaluation, and investigation, and play a key role in understanding and managing the health impacts of these viral variants.

Working with

FIND
Diagnosis for all

Ethical patient engagement

Genomics England is collaborating with precision medicine company Sano Genetics to develop a novel patient engagement platform to allow rare disease patients and their caregivers to add vital additional information about their health and wellbeing to research databases.

We were commissioned by Genomics England to lead a legal and ethical evaluation of the new online platform. Our report assessed the likely impact of the platform and how it worked for patients, concluding that it has the potential to enrich research datasets and enhance user engagement but that there should be support for patients who may be less digitally literate.

This framework could aid the development of similar platforms to improve patient involvement in medical research in the future.

"The PHG Foundation's report is an encouraging assessment that assures us the platform will help accelerate precision medicine research while ensuring that the highest levels of data security and integrity are met along the way"

Rakhi Rajani
Chief Digital Officer, Genomics England

Working with

Genomics england

Sano

"The PHG Foundation's assessment of the legality and ethics of what we're doing provides us with a robust blueprint that will be fundamental in our ongoing product development"

Dr Patrick Short
CEO, Sano Genetics

Next steps in the sequence

The National Genomic Healthcare Strategy published in 2020, *Genome UK*, set out a ten year vision for the future of genomics and personalised medicine. We welcomed the strategy, but cautioned that the details of how it was implemented would be critical for success.

The implementation plans for 2021 revealed initial commitments for the major organisations and stakeholders in genomic research and healthcare.

We hosted a digital panel event in which the then Parliamentary Under-Secretary of State of Innovation, Lord Bethell, introduced the new plans to progress the use of genomics for rare diseases, cancer and infectious diseases, and explore new opportunities for the prevention of other common conditions.

This address was followed by analysis and discussion by a panel of expert stakeholders, chaired by our Director:

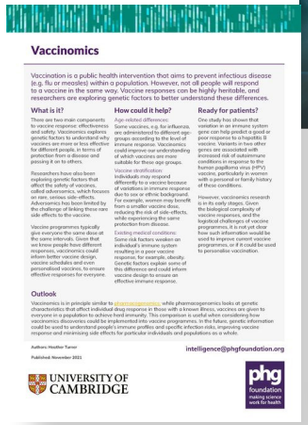
- Dr Mark Kroese (PHG Foundation)
- Chris Wigley (Genomics England)
- John Stewart (NHS England)
- Prof Anneke Lucassen (University of Southampton)
- Prof Saheer Gharbia (Public Health England)

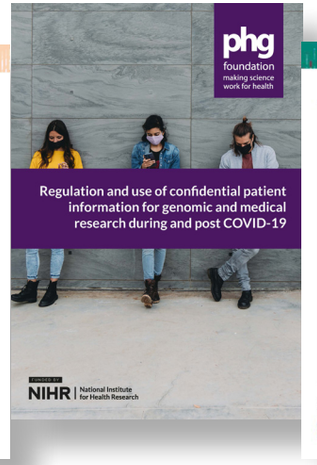
Working with

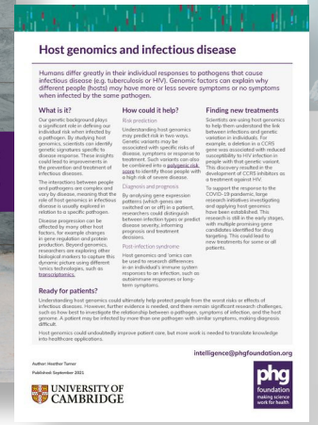
Office for Life Sciences

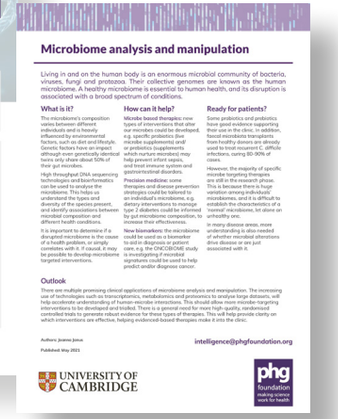
Publications

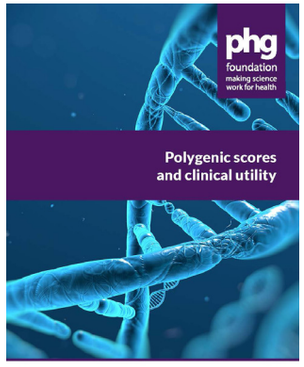
We produce knowledge products ranging from detailed reports, to shorter policy briefings, explainers and animations offering clarity on emerging technologies, opportunities and challenges. In addition, we provide updates and opinion on developments in science, health and policy via our subscriber news service and website commentary, and bring together external experts for discussion and debate. All our digital products are freely and publicly available via our website.

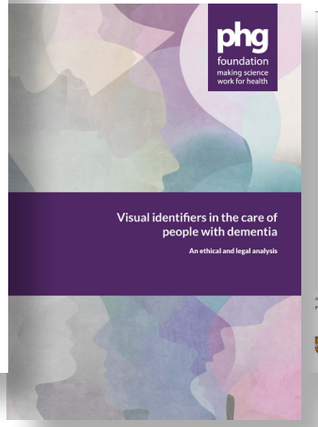


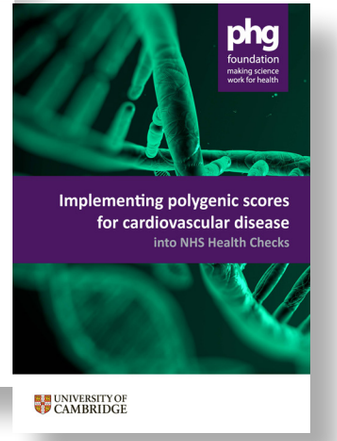






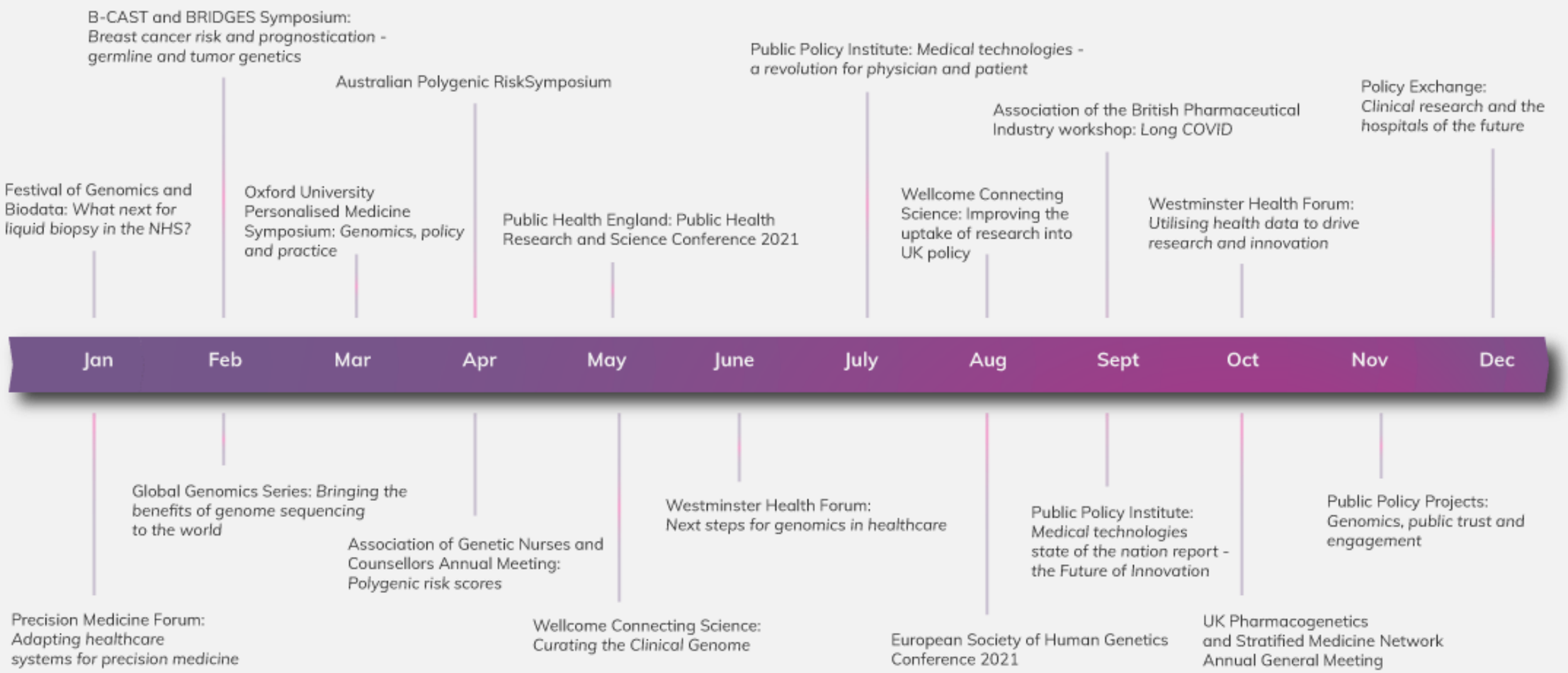






Presentations

Our senior staff represent the PHG Foundation and share our knowledge and expertise as speakers, panellists and chairs at varied events through the year. A representative selection from 2021 are shown below.



People

PHG FOUNDATION

making science work for health

Governance

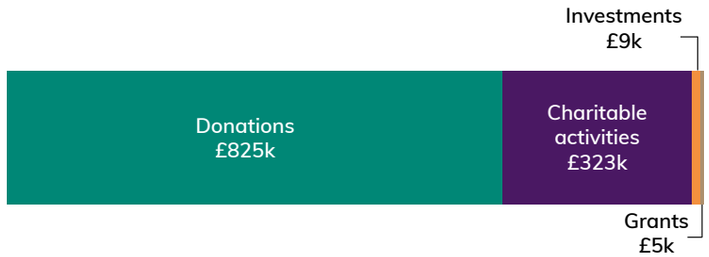
The PHG Foundation is a linked exempt charity of the University of Cambridge and its Board of Trustees comprises:

- **Prof Patrick Chinnery** - Head of the Department of Clinical Neuroscience, University of Cambridge
- **Prof Nita Ghandi Forouhi** - Professor of Population Health and Nutrition, University of Cambridge
- **Dr Anthony Freeling** - President of Hughes Hall, University of Cambridge
- **Mr Andrew Hutton** - Director, A.J. Hutton Ltd
- **Prof Patrick Maxwell** - Regius Professor of Physic, University of Cambridge
- **Prof Liba Taub** - Head of the Department of History and Philosophy of Science, University of Cambridge
- **Dr Ron Zimmern** - Founder and Chairman

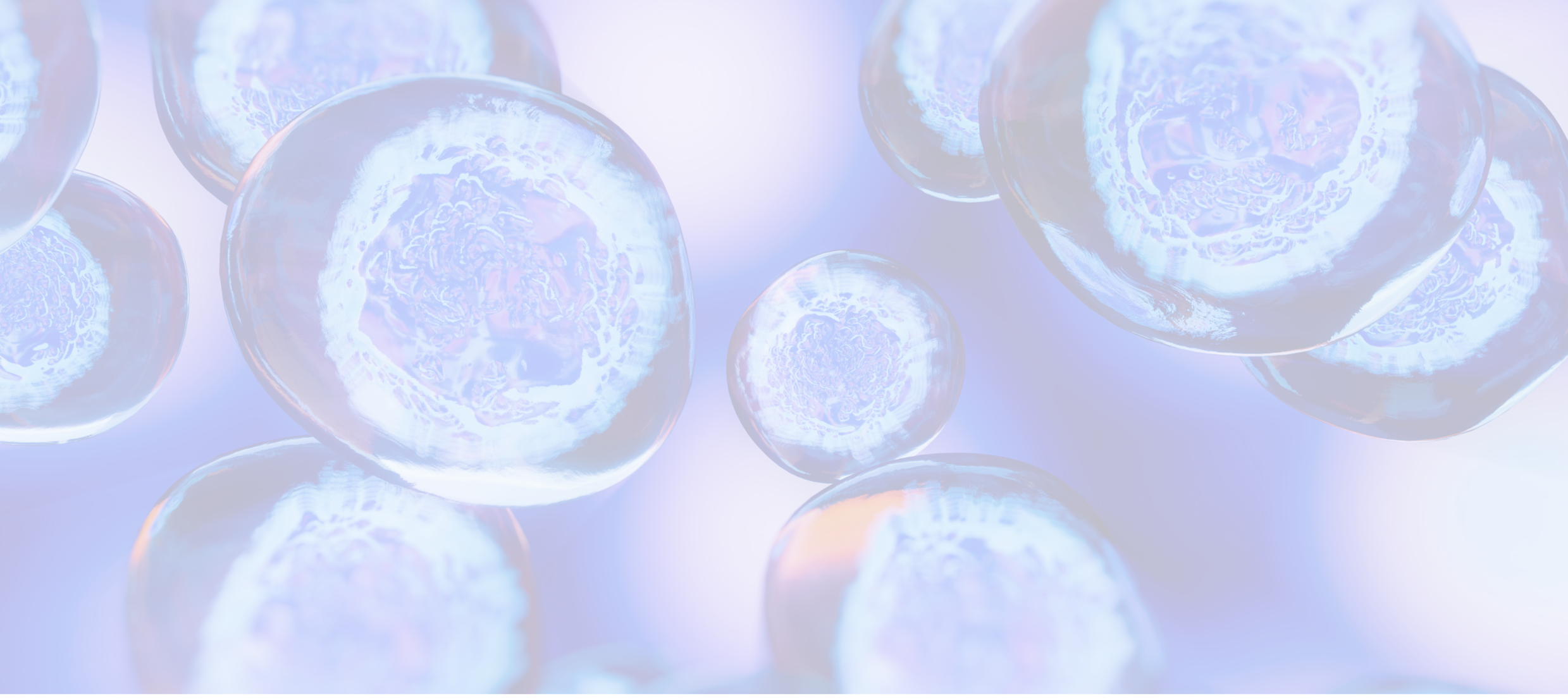
Finance

Our work is funded by philanthropic donations, primarily from the Hatton Trust and the WYNG Foundation, along with income from academic grants and collaborations, commercial and public sector commissions and consultancy, and a modest investment portfolio. We are not funded by the University of Cambridge. Most of our spending is on charitable activities, including our work programme and staff costs.

2020-2021 income: £1162k



2020-2021 outgoings: £1087k



UNIVERSITY OF CAMBRIDGE

PHG FOUNDATION

making science work for health