phg
foundation
making science
work for health

2020

UNIVERSITY OF
CAMBRIDGE

From 2020 to the future

2020 was an exceptional year for everyone; it was humbling to see the NHS, public health, social care, research and policy communities work so hard to meet the extreme challenges posed by the COVID-19 pandemic.

The last year is a dramatic demonstration of the importance of science for health. Genomics has been critical to national and global policy responses. The UK genomics community has made remarkable contributions to our understanding of the nature and behaviour of the virus, and the best ways to fight it.

I am pleased that the PHG Foundation team, working diligently and largely remotely throughout the year, has been able to continue to analyse and explain both the healthcare potential and policy issues posed by the fast-emerging science – in infectious disease genomics, and much more besides.

For now, we are still grappling with the immediate impact of the pandemic, but as we move forwards the long-term lessons and implications for healthcare must be considered. Genomics and associated technologies also offer us new answers and opportunities to protect and improve health for people and populations. The PHG Foundation will continue to bring our multi-disciplinary expertise to bear on informing and influencing relevant policy and decision-making, and helping science deliver for health.

Dr Mark Kroese
Director - PHG Foundation

Ahead of the curve

The highlight of 2020 was the release of the much-anticipated National Genomics Healthcare Strategy by the government.

The report *Genome UK: The future of healthcare* recognises the wide-ranging potential of genomics to improve healthcare for the whole population, and sets out ambitious plans to establish the most advanced genomic healthcare system in the world.

The proposed measures build on existing strengths in genomic medicine and research with a view to improving prediction, prevention, diagnosis and management of disease.

The new strategy is firmly in line with all the PHG Foundation's major policy positions on genomics, not least the call for a governmental 'policy framework for the clinical and public health applications of genetics' that we first made 20 years ago, in our Genetics Scenario Project report for the Nuffield Foundation.

Genomics goes mainstream

Over the years we have worked tirelessly alongside the genomics and policy communities to promote the potential value of genomics to improve healthcare, and to develop the steps needed to get us there. As the founding centre for public health genomics in the UK, we have always argued for making the most of opportunities to improve population as well as individual health.

Now at last, with a national NHS Genomic Medicine Service in action and new governmental plans and commitments to take advantage of developments in genomics in place, we are delighted to see this long-imagined future becoming reality.

Genomics has the potential to transform the future of healthcare by offering patients the very best predictive, preventative and personalised care

Matt Hancock
Secretary of State for Health and Social Care

Pillars of genomics

The new genomic healthcare strategy is founded on three 'pillars', or areas of focus.

The first is on improving diagnosis, stratification and treatment of disease. It builds on the existing NHS Genomic Medicine Service, including provisions for whole genome sequencing in clinical care, the use of genomic and related data for cancer care, and pharmacogenomics. It covers many of the areas we have previously highlighted for policy attention, such as ctDNA for cancer care, and innovative techniques such as long-read sequencing.

The second pillar covers the integration of biomedical research with clinical care and development. The strategy makes welcome promises to provide robust infrastructure, standards and protocols for storing and sharing genomic data, and promises to increase the ethnic diversity of datasets. This matches our 2015 call for 'NHS systems for comprehensive data collection, curation and access for clinical purposes, including a national genomic database'.

The third pillar emphasises the use of genomics to enable predictive and preventive care – that is, public health genomics. As the founding UK centre for this very field, the PHG Foundation is delighted to see the commitments to making the most of opportunities for genomics to benefit population health. Targeted (personalised or risk stratified) screening is an area we've been working in for many years, but as evidence of utility grows, there is increasing need for a policy focus to address the obstacles to effective use.

This also covers the use of genomics for control of infectious diseases. We first highlighted the potential of pathogen genomics for public health back in 2015, and the COVID-19 pandemic has since demonstrated its value to the whole world. The government also plans to develop the use of genomics in understanding individual responses to infection and how this could inform risk assessment and health protection.

We must take advantage of developments in genomics to increase prevention and early diagnosis of disease

Lord Bethell
Minister for Innovation

Promoting **personalised** medicine

As part of a European Commission funded research project, Breast Cancer Stratification (B-CAST), we have led work examining the potential for developing more personalised prevention for breast cancer based on genomic information.

Our 2020 report outlined the fast-moving science behind breast cancer prevention and made recommendations for national health systems to bridge the gap between research and policy in accelerating delivery of more accurate risk-based prevention.

Following our 2018 report on personalised medicine for NHS England, the AHSN Network commissioned us to research and produce a new report on five pivotal 'omic technologies rapidly transforming the landscape.

Genomic innovation: technologies for personalised medicine outlined the current status of pharmacogenomics, transcriptomics, circulating tumour DNA for non-invasive testing, genetically-modified regenerative medicines and technologies for near-patient testing to support antimicrobial stewardship. We also provided recommendations for NHS implementation of each technology.

This report looks at the unique ways that the 'omics era can help define our future healthcare

Prof Sir Mark Caulfield
Chief Scientist - Genomics England

A risky business?

Genomics undoubtedly contributes to our personal risk of developing disease, and could offer improved risk prediction and disease prevention.

This could apply to both infectious diseases and to common, complex conditions such as cancer and heart disease, where better understanding of individual risk could help deliver personalised approaches to prevention.

The main barrier is that the genetic influences over most diseases are very complex. Polygenic scores combine information from many individually low-risk genetic variants into a single estimate of risk for a given condition. Our ongoing work in this fast-moving field keeps pace with the science to produce timely and relevant policy intelligence and insights.

Our work in 2020 explored the evidence and discussion around the potential use of polygenic scores in healthcare. We considered the value of tests based on polygenic scores for individuals (personal utility) and for medical decision-making (clinical utility), as well as examining wider issues that the use of such tests may pose.

Insurance implications

Through a University of Cambridge partnership, we produced a report for the Data Ethics Forum of a major global insurance company.

Personalised Medicine and Insurance examines the changing obligations of insurers to their customers and wider society that may arise from increasingly tailored health risk assessments, with particular reference to data ethics and regulatory issues.

Data diversity

One important concern about using genomics in the prediction and prevention of disease is the current lack of ethnic diversity in genomic databases, in which people of European ancestry are currently over-represented, largely because of the countries where such population databases were first created.

This is an issue that needs to be addressed to ensure that the use of genomic risk assessment tests (and other forms of genomic medicine) do not worsen existing health disparities. Polygenic risk scores calculated based on data from a population group of limited diversity are less likely to work well in individuals of different ethnicity; this is an area of ongoing research.

Dealing with data

Sharing genomic data is a vital part of both medical care and research, but how do we balance that with the need to protect peoples' personal data?

The genomics community is increasingly concerned about how data protection legislation governs the use of genomic information, and what this means in practice – and the answers are far from clear.

With funding from the Information Commissioner's Office, PHG Foundation examined the impact of the EU GDPR and UK Data Protection Act on genomic healthcare and research.

We asked how far genetic and genomic data used for healthcare and medical research count as 'personal data' in law; what this may mean for how data is used in clinical research and practice in the years ahead, and what can be done to protect critical data use.

Our legal analysis found that the situation is indeed highly complex. The legislation is likely to have substantial and serious impacts for genomic medicine. These include difficulties in understanding how legal principles and rights should be applied to genomic data and challenges for international data sharing. The main problem lies in providing appropriate guidance about how very broad legal requirements should be applied in the very specific context of genomics.

We have highlighted areas where urgent action is needed to address the most serious problems, making practical recommendations for regulators, researchers and health services and calling for close collaboration between experts in genomic medicine, research and data protection at national and international levels to find solutions.

Achieving consensus for key aspects of genomic data processing could improve harmonisation and offer legal certainty

Colin Mitchell
Senior policy analyst - PHG Foundation

Lessons from a pandemic

Thanks to public and private sector commissions, our science team completed some significant research examining the field of genomic epidemiology, and the ways in which current and emerging tools and technologies for DNA sequencing and analysis can be used to detect and manage infectious diseases.

Always a fast-moving field, throughout 2020 the science base moved at breakneck speed. New evidence from COVID-19 sequencing studies around the world provided a rich resource for examination, but our work also considered the application of new sequencing approaches in different contexts, and for other potential public health threats, including viral haemorrhagic fevers such as Ebola, Lassa fever and yellow fever.

Throughout 2020 there was increasing demand for PHG Foundation's ability to unpack and explain the role of science and technology in infectious diseases, aided by global media coverage of our pre-pandemic briefing on RNA vaccines. This led us to produce a range of articles and materials on new scientific findings or applications relevant to COVID-19, including a new animated explainer about pandemic genomics.

We also contributed to a range of external policy discussions about healthcare issues raised by the pandemic, including how to build on experiences of rapid, responsive oversight and regulatory processes for the swifter movement of science into healthcare in the future. Our legal experts helped prepare guidance on responsible data sharing for COVID-19 pandemic responses for the Global Alliance for Genomics and Health.

For the first time, genomic sequencing can help to guide the public response to a pandemic

World Health Organization
January 2021

Healthcare horizons

Just how do genes do what they do? Decoding the genome was just the first step towards answering this fundamental question; there's a great deal we still don't understand. Functional genomics is a field that uses multiple 'omics datasets from different levels of biological activity to understand the complex relationship between our genes and their visible effects. It could provide valuable insights into the implications of genetic variation for health and disease.

Our policy briefings on this topic outline what functional genomics is and how it could be used to improve clinical care. In the short term, insights from this field could help diagnose rare diseases and improve understanding of cancer; ultimately, it could support delivery of more personalised medicine for common diseases.

Looking ahead to the technologies of the future is an important part of our work, helping us to anticipate healthcare impacts and policy implications. We have an ongoing programme of appraisal, investigating the science and status of technologies for health in order to cut through the hype and provide a clear assessment of potential.

We look at the literature and talk to experts to make sense of research directions and applications, and understand both how technologies are being used today, and where they might impact healthcare tomorrow.

Whilst much of our horizon scanning goes on in the background, we also share some of our findings in the form of briefings and explainers on different topics.

Science constantly throws up exciting new opportunities for health - the challenge is finding those that may have the biggest impact and understanding the policy issues

Laura Blackburn
Head of Science - PHG Foundation

Citizen data *for* health

Governments around the world are interested in the risks and rewards of tapping into the growing wealth of digital information produced by people, often unconsciously - citizen generated data (CGD). Whilst there is already policy debate around the economic and social implications of harnessing CGD, there has been relatively little consideration of how CGD could affect health.

Health systems are grappling with how to make the best use of the many forms of patient data they generate, but could CGD produced outside healthcare settings also offer new opportunities to improve health? Data collected throughout the life-course by people who are healthy, unaware of increased disease risk, or showing early signs of ill health are a source of potentially useful information to inform prevention strategies, both for individuals and populations. Such a course is not without risks, and the potential benefits must be balanced with careful consideration of threats including privacy and confidentiality.

PHG Foundation examined the prospects for using CGD for health, and specifically to support predictive prevention of disease – more personalised assessment of risk and targeted interventions to keep people well for longer, and to reduce the burden on health systems.

We found that there is a wealth of data that could inform better prevention, ranging from information generated by health apps and personal medical devices to that from online shopping and searches, smartphone use and even digital banking. To make the best of this opportunity, health services have to be open to using this data – and that would mean systems and standards for easy data integration, careful consideration of the potential risks, and engagement with citizens to build trust and understanding.

We could make much better use of citizen data to improve health - but we need to work closely with the public

Philippa Brice
External Affairs Director - PHG Foundation

Learning health machines

Modern healthcare is increasingly based on digital data, and the more complex the data, the more important artificial intelligence (AI) techniques, notably machine learning and deep learning, become to make sense of it. But these new and fast-evolving approaches also pose new policy problems.

AI could revolutionise genomic medicine, streamlining the analysis of huge volumes of DNA sequence information and integrating it with other related sources of data to enable rapid, effective analysis.

PHG Foundation examined how AI offers potential improvements across the whole genomic data pipeline – and the limitations that must be addressed to realise this potential. Informed by expert consultation, we set out the priority policy actions, ranging from skills and infrastructure needs through to measures to reduce bias and promote cross-disciplinary collaborations.

Unpacking the black box

AI has many applications in healthcare, not least for earlier and more precise disease diagnosis and public health surveillance of disease risks. Computerised analysis of medical data by opaque or 'black box' machine learning processes can outperform even expert professionals, but the logic by which these models reach conclusions is not obvious. This poses novel challenges for proper regulation, and could also erode trust.

Funded by the Wellcome Trust, our Black box medicine and transparency project examined what ethical and legal rules should, and could, apply to 'black box' medicine. The PHG Foundation research produced guidance and tools for legal, regulatory and health policy audiences, and for developers of machine learning models for healthcare, to help them find the right balance between performance and protection in different contexts.

AI could transform healthcare, but the opaque and dynamic nature of some AI models raises significant regulatory challenges

Alison Hall
Head of Humanities - PHG Foundation

Science powered policy

Before the pandemic restrictions took shape, we were privileged to host a lively discussion event, *Creating policy impact - international lessons from science and health.*

Our speakers were Prof Eric Meslin, President of the Council of Canadian Academies, which provides independent expert science advice to inform public decision-making; and Prof Dame Sally Davies, UK Special Envoy on Antimicrobial Resistance and former Chief Medical Officer for England.

Having advised presidents and prime ministers, and informed national and international policy deliberations, our guests were able to provide all kinds of valuable insights and anecdotes on how, when and where to use evidence to influence policy – though none of us realised that the subject of science advice to governments would soon be hitting the headlines on a daily basis.

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The switch to remote interactions in March 2020 did not prevent our staff from speaking at a wide range of meetings to share ideas and intelligence

CRUK
-
Translating genomics in oncology

HKU
-
Health and AI: law, ethics and society

New Statesman
-
Age of the Genome

COVID Commission
-
Policy seminar

Westminster Forum
-
Prevention and public health

MedTech Summit 2020
-
Software Design for Medical Devices 2020

ExACT
-
Personalising breast cancer prevention

Hughes Hall
-
The future of ageing

Royal Society of Medicine
-
Personalised care for cancer

Health Service Journal
-
Precision medicine roundtable

16th World Congress on Public Health
-
Opportunities and challenges for PRS

AHSN Network
-
Technologies for personalised medicine

PHE
-
CVD prevention conference

Finances

As a linked exempt charity of the University of Cambridge, much of our funding comes from generous donations from the Hatton Trust and the WYNG Foundation. We are not funded by the University of Cambridge, but we do receive income from academic grants and collaborations, commissions and consultancy from public, commercial and third sector partners, and a modest investment portfolio.

2019-2020 income: £1.15m

Charitable activities: £122k

Other: £42

Donations: £832k

Investments: £25k

Grants: £161k
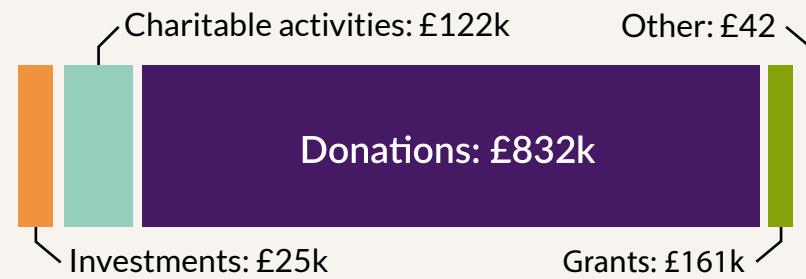
2019-2020 outgoings: £1.21m

Fundraising £2k

Support: £117k

Charitable activities: £1.07m

Governance: £26k

Most of our spending goes on our charitable activities, with a little extra on the essentials; 2019-20 saw a slight planned overspend, taken from our financial reserves.