



HM Government

# **Genome UK:**

**2022 to 2025**

**Implementation plan for England**



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## Ministerial foreword

Our country leads the world in genomic healthcare and research. This has never been more evident than in the last 2 years, when we were at the forefront of monitoring and tracking variants of the COVID-19 virus. The NHS in England is the first healthcare service in the world to offer whole genome sequencing as a part of routine care, and we hold unique research resources such as UK Biobank.

In 2020 we published our [Genome UK: the future of healthcare](#) strategy, which outlines how we will become the most advanced genomic healthcare system in the world. Earlier this year, we published our [shared commitments for UK-wide implementation 2022 to 2025](#), setting out an ambitious plan to ensure genomics research and healthcare can flourish across the whole of the UK.

We are now publishing an implementation plan for England, which builds on the shared commitments and outlines the actions we will take in England over the next 3 years. This plan will take us to the halfway mark of the original strategy in 2025, which means that making strong progress in these 3 years will be pivotal to delivering our ambitions.

Our delivery partners will take forward a world-leading programme of innovation in genomics diagnostics and clinical services, evaluation of new genomic tools in prevention and early detection of disease, and cutting-edge genomics research, all enabled by new, large-scale data capabilities.

The delivery of genomic healthcare for patients through the NHS is continuing to develop at pace too. Genomics England and NHS England will work together to bring innovative genomic technologies closer to the patient, with the evaluation of new sequencing technology to improve the accuracy and speed of diagnosis for cancer patients. NHS England will continue the annual review of the National Genomic Test Directory so that more patients are eligible for genomic testing and, through the NHS Genomic Medicine Service (GMS) Alliance transformation projects, will test new diagnostic technologies to ensure that the most innovative and effective genomic technologies are available for use in patient care. Taken together, these actions will deliver real improvements in patient care and health outcomes.

To start this important next phase of our implementation work, the government is announcing the following investments:

- ▶ £105 million for a landmark research programme, led by Genomics England in partnership with the NHS, to study the effectiveness of using whole genome sequencing to speed up diagnosis and treatment of rare genetic diseases in newborns, potentially leading to life-saving interventions for thousands of babies
- ▶ £22 million for Genomics England to tackle health inequalities in genomic medicine through tailored sequencing of 15,000 to 25,000 participants from diverse backgrounds by 2025, as well as extensive community engagement work to build trusted relationships with traditionally excluded groups of people
- ▶ £26 million for an innovative cancer programme, led by Genomics England in partnership with NHS England and the National Pathology Imaging Co-operative, to evaluate cutting-edge genomic sequencing technology and use artificial intelligence to analyse genomic data alongside digital histopathology and radiology images, improving the accuracy and speed of diagnosis for cancer patients

- ▶ up to £25 million Medical Research Council-led funding for a 4-year functional genomics initiative, working across UK Research and Innovation and other stakeholders to establish an industry-partnered world-class offer on functional genomics, building on already existing infrastructure and UK research expertise

These initiatives will join existing programmes across the UK already, including within Genomics England and the NHS GMS. Our Future Health plans to genotype the world's largest population cohort to support the early detection of disease, and UK Biobank is continuing work to maximise the capabilities of its resource – the world's most characterised and widely used research cohort.

The UK has the opportunity to be a genomic superpower. We are already seeing the results of the innovations in genomic healthcare and research in the UK, revolutionising outcomes for patients and generating valuable new investments. This plan will drive this transformation forward.



**Will Quince**

Minister of State (Minister of State for Health), DHSC



**Nusrat Ghani**

Minister of State (Minister for Industry and Investment Security), BEIS

## Executive summary

The UK is a global leader in genetics and genomics. To maintain and extend this leadership position, the UK government published '[Genome UK: the future of healthcare](#)' in September 2020 – a 10-year strategy to create the most advanced genomic healthcare system in the world and deliver better health outcomes at lower cost. In March 2022, the UK government and devolved governments published '[Genome UK: shared commitments for UK-wide implementation 2022 to 2025](#)', setting out how they will work together to implement the Genome UK strategy. Recognising the devolved responsibilities, we agreed that the UK government and the devolved governments would each publish separate, nation-specific plans.

This implementation plan for England lays out specific actions that our genomics delivery partners in England will take during the 2022 to 2025 spending review period to implement the commitments in Genome UK. The plan showcases the outstanding research and policy work that is taking place to develop, evaluate and implement new genomic technologies. The actions are not exhaustive – the UK has a very active genomics research and clinical service community, and it would not be possible to include every project, pilot study or trial. We have focussed on the key projects put forward by our delivery partners, while recognising that this only represents some of the excellent work underway across England.

Successful delivery of the implementation actions described here will ensure that by 2025, we will have made significant progress in realising the benefits of genomic healthcare. Genomic technologies that support early detection of disease, enable faster and more accurate diagnoses, and speed up the development of personalised, more effective treatments are important tools that will deliver better health outcomes for patients and support sustainable delivery of our healthcare system. We expect that by 2025, genomic healthcare will play a significant role in enabling healthcare reform, propelled by a growing genomics research sector which in turn will play an important part in generating and supporting economic growth.

For the first time, we will also publish a suite of metrics which will determine the combined impact of the initiatives included in this plan and measure progress against our Genome UK objectives.

### Summary of key actions for the next implementation period of Genome UK

We will progress our ambitions for diagnosis and personalised medicine, incorporating the latest genomics advances into routine healthcare to improve the diagnosis, stratification and treatment of illness. Our implementation plan includes the following actions:

- ▶ Genomics England, in partnership with NHS England and the National Pathology Imaging Co-operative, will lead the Cancer 2.0 programme – a £26 million innovative cancer programme to evaluate cutting-edge genomic sequencing technology. The programme will use artificial intelligence to analyse genomic data alongside digital histopathology and radiology images to improve the accuracy and speed of diagnosis for cancer patients.
- ▶ NHS England will continue to introduce new clinical indications for genomic testing through the National Genomic Test Directory so that more patients can benefit from genomic testing. NHS England will also pilot innovative technologies through the NHS GMS Alliance transformation projects to build the evidence base for adopting more genomic technologies in the future.

- ▶ Genomics England and NHS England will deliver the Diagnostic Discovery pathway, enabling the discovery of new diagnoses for patients with rare disease using the latest research findings. This provides benefits to individual families and insights to inform future development of the NHS GMS, enabling even more rare disease patients to receive a diagnosis in the future.

We will progress our ambitions for effective prevention and early detection, generating evidence about the utility of new genomic tests and using genomic insight to improve health outcomes and transform the delivery of healthcare. Our implementation plan includes the following actions:

- ▶ Genomics England will lead on delivering £105 million investment in a landmark research programme, in partnership with the NHS, to study the effectiveness of using whole genome sequencing to speed up diagnosis and treatment of rare genetic diseases in newborns, potentially leading to thousands of life-saving interventions.
- ▶ UK Biobank will build on its strength as the largest collection of whole genome sequencing (WGS) data in the world, and develop a secure and scalable genome variant imputation service to enrich data collected by Our Future Health and other resources using lower cost genotyping, further improving UK genomic resources for genomic analysis.
- ▶ Our Future Health will recruit up to 5 million participants by the end of 2025, making it the UK's largest ever health research programme. In partnership with Genomics plc, Our Future Health plans to calculate polygenic risk scores (PRS) and offer participants healthcare insights, should they choose to receive them. These activities and the uniquely large cohort will enable the research community to advance our understanding of disease risk and build the evidence base on the utility of PRS in healthcare.

We will progress our ambitions to extend our lead in genomic research and data. We will support the continuous development of cutting-edge genomic technology, move towards a future federated ecosystem by improved data standards and interoperability of genomic datasets for research use, and improve patient access to clinical trials. Our implementation plan includes the following actions:

- ▶ The Medical Research Council will provide up to £25 million investment in a 4-year functional genomics initiative, working across UK Research and Innovation and other stakeholders. This initiative will respond to views from UK researchers and industry partners about the priorities for establishing a world-class offer on functional genomics, building on already existing infrastructure and UK research expertise. Open competition funding mechanisms are expected to launch in spring 2023.
- ▶ NHS England will continue to develop unrivalled at-scale data infrastructure (secure data environments) to deliver key research and development opportunities, making a variety of data types available in a streamlined, secure and privacy-protected way. This includes working with the NHS to make more clinical test sequences available for research use.
- ▶ Our Future Health will use genotype array and other genomic data from its 5 million diverse population cohort to support the development and evaluation of genomic tools in prevention and early disease detection. The genomic data will be combined with questionnaire and health-related data to create a new research platform which is unique in cohort size and supports discovery and translational research at scale. Participants can consent to be recontacted, enabling them to receive health-related insights and be invited to additional studies based on disease risks.

- ▶ UK Biobank, one of the world's most genetically characterised and used research resources, will use £20 million funding from the Wellcome Trust to develop its research analysis platform, further developing cloud-based platform functionality and making the platform even more accessible with a wide range of analytical tools. By end of 2023, all genomic data will be available to industry partners and approved researchers for analysis alongside over 15 years of follow-up health outcomes data, enabling in-depth research into the genetic determinants of disease.
- ▶ UK Biobank will use £30 million funding by the Medical Research Council, Calico Life Sciences and Chan Zuckerberg Initiative for repeat magnetic resonance imaging of up to 6,000 UK Biobank participants over the next 6 years, creating the world's largest longitudinal imaging dataset.
- ▶ Genomics England will lead a £22 million programme to carry out tailored genomic sequencing of 15,000 to 25,000 research participants from diverse ancestry groups that are currently under-represented in genomic research. This will increase our understanding of genomic diversity and its impact on scientific, clinical and health system outcomes, aiming to reduce health inequalities, and improve patient outcomes across all communities.
- ▶ The National Institute for Health and Care Research (NIHR) BioResource will use £40 million of NIHR funding to continue to build and enhance phenotyping of disease cohorts, including rare diseases, and increase the inclusivity and diversity of existing cohorts. This work will contribute to the long-term goal of creating a national infrastructure platform to enable the rapid recruitment to clinical trials and studies, including better identification of suitable participants.



# Introduction

In September 2020, the UK government published '[Genome UK – the future of healthcare](#)', a 10-year strategy to create the most advanced genomic healthcare system in the world, delivering better patient outcomes at lower cost. It also positions the UK to become the best location globally for genomics research and investment to grow new genomics healthcare companies.

Our aim is to ensure that people and patients across the UK can benefit fully from genomic healthcare through a more preventative approach, faster diagnosis and more personalised treatment leading to improved long-term outcomes. Researchers and industry will be supported in their research and incentivised to secure the UK's position as an international leader in genomics.

We have adopted a phased approach to implementing the strategy, allowing us to reflect emerging science. In May 2021, we published the [2021 to 2022 Implementation Plan](#) for Genome UK, setting out priority actions for the financial year 2021/22 in England, with contributions from the Scottish and Welsh governments.

In its recent report '[Harnessing the UK's genomics expertise to improve patient outcomes](#)', the Association of the British Pharmaceutical Industry (ABPI) highlighted the importance of working across the four nations of the UK to achieving our Genome UK ambitions.

While recognising that the planning and delivery of healthcare, including genomic healthcare, is devolved, we agree that concerted, UK-wide action is crucial. There is a clear benefit in working together to progress our common ambition of improving patient care and growing the sector. This is why, in March 2022, we published – jointly with the devolved governments – '[Genome UK: Shared Commitments for UK-wide implementation 2022 to 2025](#)', which sets out our joint commitments for better UK-wide co-ordination and collaboration in genomic research and healthcare.

The 2021 spending review, which set departmental budgets in England and devolved government allocations for 2022 to 2025, provided a timely opportunity to set out how we will progress implementation of the Genome UK vision over the next 3 years in England, working with delivery partners. In 2025 we will be marking the half-way milestone in the Genome UK 10-year timescale, and measurable progress in the next 3 years will be critical to successfully delivering the vision set out in the strategy.

## The UK genomics landscape

The UK has a vibrant genomics research and healthcare landscape bringing together the NHS, world-leading research assets and a thriving life sciences sector.

Following the completion of the 100,000 Genomes Project in 2018, Genomics England – a Department of Health and Social Care-owned company – holds the largest global research collection of whole genome sequences from patients with cancer and rare diseases. This number is increasing with de-identified genomic data from the NHS GMS being transferred to the Genomics England National Genomic Research Library, a secure national research environment of genomic and health data, with patient consent.

UK Biobank, established in 2006, has sequenced the exomes and whole genomes of its 500,000 participants. It now represents the largest collection of genome sequences anywhere in the world, all of which are linked to participants' detailed NHS health records. Both UK Biobank and Genomics England are now also linking imaging data to already available clinical and genomic datasets.

In 2019, the government established [Our Future Health](#) (formerly known as the Accelerating Detection of Disease Challenge) with £79 million from the Industrial Strategy Challenge Fund, via Innovate UK. With volunteers' consent, Our Future Health will aim to collect and link multiple sources of health and lifestyle information, including genetic data, across a cohort of 5 million adults that truly reflect the UK population. Data will be held in a secure data store and will be deidentified before being made available for health research in trusted research environments. This will create a unique research platform enabling both discovery and translational research at scale, as well as allowing researchers to re-contact and invite participants to future studies based on their genetic and health information. Our Future Health's planned research programme has already attracted £150 million of industry investment and is expected to make a significant contribution across all Genome UK objectives, with particular benefits expected in disease detection – an important strategic goal which will result in better outcomes for patients. Our Future Health is a UK wide organisation which is expected to recruit participants across the 4 nations. The recruitment of volunteers and data collection has already begun in England, and Our Future Health will continue to work with the devolved governments to develop and roll out plans for Scotland, Wales and Northern Ireland, including recruitment initiatives, logistics and infrastructure building.

In October 2022, NHS England published the first ever NHS genomics strategy '[Accelerating genomic medicine in the NHS](#)', which sets the strategic direction and priorities of its genomics programme over the next 5 years. The strategy aims to ensure that genomics will be at the heart of a sustainable NHS in the future and the next generation of healthcare in the NHS. It sets out 4 priority areas to this approach:

- ▶ embedding genomics in the NHS through a world leading, innovative service model
- ▶ delivering equitable genomic testing for improved prediction, prevention, diagnosis and precision medicine
- ▶ enabling genomics to be at the forefront of the data and digital revolution
- ▶ evolving the service through cutting-edge science, research and innovation

The life sciences sector offers important and unique opportunities for generating investment and supporting economic growth across the UK. We know that the sector provides a huge boost to the UK economy, generating a turnover of £94.2 billion in 2021, and employing 282,000 people across the UK. [65% of these jobs are outside of London and the South East. The value of estimated inward life sciences foreign direct investment in the UK was £1.9 billion in 2021, coming behind only the USA in terms of value.](#)

A range of evidence suggests that the genomics sector in particular offers high growth potential. The BioIndustry Association's report, '[Genomics Nation 2021](#)', describes the size and growth of the genomics industry in recent years and projections for growth going forward. The report estimates that while the sector's market capitalisation was less than £10 billion in 2021, their forecast suggests this will reach over £50 billion by 2040. The latest publication from the Office for Life Sciences (OLS)

on the [bioscience and health technology sector statistics](#) also monitors trends in the number of businesses conducting genomics-related activity, the number of employees and the turnover generated from these sites. The latest statistics show that employment has seen substantial increases since 2009 with sharp increases year-on-year since 2017. There was a 12% rise in employment at sites with genomics activity between 2020 and 2021.

## **UK genomics governance**

The [National Genomics Board](#), which brings together senior decision makers and representatives from across the genomics sector, including senior officials from the devolved governments, provides strategic oversight and works collaboratively across the UK to harness the benefits of genomic healthcare – ultimately helping to ensure delivery of the vision set out in Genome UK. The National Genomics Board is chaired by the minister responsible for life sciences in the Department of Health and Social Care.

To monitor more detailed progress on Genome UK implementation plans, OLS leads an implementation co-ordination group. The group convenes delivery leads for programmes and projects in England under the pillars and themes set out in the Genome UK strategy and provides a forum for discussion on coordination and progress of implementation commitments and actions included in implementation plans. The implementation co-ordination group also provides specific updates on progress relating to implementation actions to the National Genomics Board, as appropriate.

Together with the devolved governments, OLS has also set up a UK shared commitments group, attended by representatives from the health services in the 4 governments and chaired by OLS. The group considers high-level co-ordination and progress in delivery of the UK-wide shared commitments, as well as agenda items for the National Genomics Board.

## **Measuring the impact of Genome UK implementation**

To assess the long-term impact of implementation actions and initiatives, OLS has derived a set of high-level metrics that will quantify long-term changes in the genomics environment and measure progress against Genome UK ambitions. These metrics have been discussed and agreed with delivery partners and are outlined in Annex A, published alongside this plan. OLS will undertake further work to refine or expand the list of metrics based on evolving data availability and feedback received following the publication of the implementation plan.

These metrics relate to England and we are working with the devolved governments to achieve a harmonised approach across the UK where possible.

We will publish further information via a metrics baseline report in due course. This will include:

- ▶ further specification on the metrics, including scope and definitions
- ▶ baseline figures for each metric that quantify the situation at the earliest possible timepoint
- ▶ any additional metrics that are important for measuring the impact of implementation actions

We plan to publish future implementation progress reports and appropriate updates on metrics.

# Implementation actions 2022 to 2025

## Diagnosis and personalised medicine

As we learn more about the role and function of the genome in disease, the application of genomic technologies in diagnosis and personalised medicine is becoming even more important and impactful. Using genomics, we can provide rare disease and cancer patients with an accurate diagnosis earlier and more quickly, whilst also supporting the use and development of personalised and stratified treatments. Furthermore, extending the use of pharmacogenomics in the NHS will ensure more patients get the right treatments at the right time and at the right dose, improving outcomes for patients and reducing the number of adverse drug reactions and their impact on the NHS. Finally, using genomics can help us to better understand pathogens and how they are spread, as well as the role of a person's genome in their response to infectious diseases. This supports scientists in controlling outbreaks, as well as developing new diagnostics and treatments for infectious diseases.

In [Genome UK](#), we set out the following commitments for the 'diagnosis and personalised medicine' pillar, to:

1. ensure the NHS is ready to evaluate and implement all clinically relevant, genomic technologies and novel genomic healthcare applications based on the latest, robust evidence from experts at the forefront of their fields across the UK and globally
2. offer all patients with a rare genetic disorder a definitive molecular diagnosis using tests that will support research into their condition wherever possible
3. offer genomic testing to all people with cancer for whom it would be of clinical benefit
4. support the join up of the NHS and research community with scalable and secure informatics systems, both for clinical decision support and large-scale data processing and analytics
5. secure the best value per clinical whole genome sequence anywhere in the world, and help ensure that new clinically relevant technologies become more widely available at a competitive price
6. have a clear evidence-based position on whether and how pharmacogenomics should be implemented in the health service at scale
7. sequence pathogens quickly and easily using point of care sequencing technology, helping us control outbreaks and fight antimicrobial resistance
8. understand the role of the genome in differing patient outcomes from infectious disease
9. rapidly utilise advances in sequencing technology to develop and deploy new diagnostics and support better, more integrated, surveillance of infectious diseases
10. provide international leadership in supporting the development of best practice in infectious disease genomics and public health, through international projects such as the Global Alliance for Genomic Health and the Public Health Alliance for Genomic Epidemiology

## Actions our genomics delivery partners have taken over the past 18 months

- ▶ The NHS GMS in partnership with Genomics England increased the number of rare disease and cancer patients who can access WGS, which is a world-leading diagnostic test. As of the end of October 2022, around 33,000 whole genome equivalents have been sequenced through this service, with an average diagnostic yield of 32%, rising to up to 61% in some conditions. To ramp up the service, NHS England has made it easier for clinicians to order WGS. In April 2022 they made 20 additional clinical indications available, resulting in a total of 190 clinical indications currently available via WGS (inclusive of pilot initiatives).
- ▶ NHS England have increased the number of genomic tests available to patients, via the Test Directory. As of October 2022, the Test Directory includes 357 rare disease clinical indications, covering around 3,200 rare and inherited diseases and 203 cancer clinical indications. The Test Directory supports the NHS GMS to carry out over 680,000 genomic tests in England every year for common and rare and inherited disease, pharmacogenomics, and cancer.
- ▶ NHS England delivered a national rapid whole exome sequencing service, now a rapid WGS service, for acutely unwell children with a likely monogenic disorder (disorders likely to be caused by a defect in a single gene) in neonatal intensive care units and paediatric intensive care units. There have been around 2,500 referrals to date, with a diagnostic yield of around 40%. A genetic diagnosis can often guide the children's clinical management and treatment.
- ▶ The NHS GMS launched a world-leading National Foetal Exome Sequencing Service in October 2020, with 250 referrals to date and a diagnosis identified in around 40% of cases. This testing provides results within a rapid turnaround time to provide a diagnosis and urgently inform clinical management of the index pregnancy.
- ▶ The UK Health Security Agency expanded COVID-19 viral sequencing capacity within the UK to support national research studies, assessment of vaccine efficacy and evaluation of diagnostic or variants detection testing against genome sequences.
- ▶ The UK Health Security Agency began building a public health service infrastructure for pathogen genomics with regional and national sequencing hubs across the UK to support SARS-CoV-2 sequencing and to establish the foundation of a pathogen sequencing framework.
- ▶ The government provided international leadership in supporting the development of best practice in infectious disease genomics, including through the International Pathogen Surveillance Network Global Pandemic Radar, to strengthen global genomic surveillance.
- ▶ The Department of Health and Social Care published [England's Rare Diseases Action Plan](#) on 28 February 2022, setting out 16 specific, measurable actions for the next year under the 4 priority areas of the UK rare diseases framework. These include 2 actions which support the commitment in Genome UK to make progress on the roll-out of WGS to patients with a suspected rare disease.
- ▶ NHS England published their genomics strategy, '[Accelerating genomic medicine in the NHS](#)' to set the strategic direction and priorities of its genomics programme over the next 5 years.

## Actions our genomics delivery partners will take over the next 3 years

- ▶ Genomics England will deliver a £26 million ‘Cancer 2.0’ programme to improve the speed and accuracy of diagnosis for cancer patients. The programme will have 2 main components. Firstly, it will explore the use of novel, long-read WGS within a clinical setting, which has the potential to provide faster, more comprehensive and accurate diagnostic capabilities for certain cancers (compared to the short-read sequencing technology currently used by Genomics England). Secondly, it will work with the National Pathology Imaging Co-operative to combine digital histopathology and radiology images with WGS data – which will be accessible to approved researchers through Genomics England’s research environment. Genomics England will partner with researchers across industry and academia to analyse the multi-modal data at scale using machine learning technology, deriving novel insights in cancer research that enable better predictive models of diagnosis, prognosis and response to treatment. This work builds on the success of the National Pathology Imaging Co-operative, which was created as part of £100 million government investment in 2018 to establish digital pathology and imaging AI centres of excellence across the UK. The work enables new ways of using artificial intelligence to analyse medical imaging and pathology data, speeding up the diagnosis of diseases (Genome UK commitments 1, 3, 4, 24 and 28).
- ▶ NHS England is developing a genomics informatics implementation plan to outline how the NHS genomics data infrastructure will support interoperability of data and drive efficiencies across the spectrum from ordering a test through to availability of this data for research at scale. This will enable quicker results for patients and continue to support innovation in genomic healthcare. This commitment was included as one of the 4 priority areas set out in the recently published [NHS England strategy](#) (Genome UK commitments 4 and 26).
- ▶ The NHS GMS will continue to update the Test Directory annually so that more clinical indications (and therefore, more patients) are eligible for genomic testing. NHS England, supported by a genomics clinical reference group and test evaluation working groups, will review the Test Directory on an annual basis to keep pace with scientific and technological advances, while delivering value for money for the NHS. A robust and evidence-based process and policy is in place to ensure that genomic testing remains available for all patients where it would be for whom it would be of clinical benefit. This is supported by a horizon scanning process and fast stream application system to ensure the Test Directory can respond quickly to emerging developments (Genome UK commitments 1, 2, 3 and 6).
- ▶ The NHS GMS is exploring the introduction of innovative genomic sequencing techniques to improve diagnosis and treatment of patients, including cancer patients. The latest technologies are being piloted through a number of NHS GMS Alliance transformation projects to ensure the most innovative and effective genomic technologies – such as RNA sequencing, long-read sequencing and optical mapping, and liquid biopsies (circulating tumour DNA and ctDNA) – can be commissioned in the NHS, based on the latest evidence. This includes a pilot to assess the use of more comprehensive pharmacogenomic testing in clinical care to reduce the number of adverse drug reactions and improve the efficacy of drugs and patient outcomes (Genome UK commitments 1, 3, 5 and 6). The National Disease Registration Service – including the National Cancer Registration and Analysis Service and the National Congenital Anomaly and Rare Disease Registration Service – will work closely with the NHS GMS to support demand modelling and evaluation of the uptake of genomic testing in eligible patient groups across cancer, congenital anomalies and rare disease.

- ▶ The NHS Southwest Genomic Laboratory Hub has recently launched a new rapid WGS service for specific conditions to provide actionable WGS results more quickly for patients across England. This service will be provided to acutely unwell children with a likely monogenic disorder. Rapid WGS has the potential to increase the detection of diagnostic variants, offer more individuals a diagnosis, and enable more patients to access life-saving treatments. This could result in fewer days in hospital, fewer invasive procedures including major surgeries, and improvements in the NHS by reducing the need for multiple diagnostic tests (Genome UK commitments 1, 2 and 5).
- ▶ Over the next 3 years, the UK Health Security Agency will lead the ongoing COVID-19 response in England, including on new variants and supporting national recovery. They are maintaining COVID-19 horizon scanning and genomic surveillance throughout 2023/24. They are also using research studies as key surveillance tools, including the Office for National Statistics COVID-19 infection, antiviral efficacy and healthcare workers exposure studies (Genome UK commitment 9).
- ▶ The UK Health Security Agency is also working to reduce the harmful impact of Hepatitis B, Hepatitis C and HIV. They are establishing a programme of engagement with the NHS to collate HIV WGS data, implement sequencing methodology for HIV and implement HCV WGS in clinical laboratories (Genome UK commitment 8).
- ▶ The UK Health Security Agency has committed to enhance the resilience and scalability of national and local public health systems, by introducing standards and frameworks for data and services to facilitate responsiveness and flexible scope. They will strengthen their data and analytics capability by developing specialized analytical platforms for pathogens genome data analysis, beyond COVID-19 (Genome UK commitment 8).
- ▶ NHS Blood and Transplant (NHSBT) has established a programme to expand the use of genomic testing to deliver more accurate, personalised and rapid donor-recipient matching in blood transfusion, solid organ and stem cell transplantation. This in turn will reduce the risk of allo-immunisation for patients and for transplant rejection (Genome UK commitment 1).
- ▶ For transfusion, NHSBT is a founding member of an international collaboration between global blood services, industry and academia. In the next 12 months they will deliver validated genotyping technology for the identification of clinically relevant red blood cell antigens. In 2023/24 NHSBT, in collaboration with the NIHR BioResource, will complete the genotyping of 80,000 regular blood donors, supporting research and clinical trials to improve matching of blood products for patients with sickle cell disease and other inherited haemoglobinopathies. This work will inform the future deployment of genotyping technology in the testing and provision of more personalised blood products to benefit patients.
- ▶ For stem cells and solid organ transplantation, NHSBT has established a 3-year collaboration project with Oxford Nanopore Technologies to develop a validated rapid full HLA gene sequencing to support matching in stem cell and organ transplantation (2022/23 to 2025/26). The project aims to develop faster, more accurate and scalable sequencing which can significantly improve pathways to transplantation for patients on the organ transplant waiting list, and for patients needing stem cell transplants to treat cancer or rare diseases.

- ▶ Our Future Health is working in collaboration with NHSBT to recruit blood donors who may provide consent for Our Future Health to share genotype data with NHSBT. This data will be used as an initial screen for donors who may have rare blood types, which can then be verified by NHSBT to improve matching in blood transfusion services.
- ▶ Genomics England will enable ongoing discovery of new diagnoses for rare disease patients through the latest research developments. Research performed using data stored in the National Genomic Research Library has identified more than 1,400 new diagnoses as of September 2022. Genomics England, with NHS England, have established the diagnostic discovery pathway to return these findings efficiently to local NHS clinical teams for clinical interpretation and reporting. The findings from diagnostic discovery not only benefit individual families, but also provide insights that can inform future developments in the NHS GMS (Genome UK commitment 2). We will be able to track genomically-confirmed rare disease diagnoses through the National Congenital Anomaly and Rare Disease Registration Service.

### **Prevention and early detection**

Effective prevention, including screening and early detection of disease, has the potential to dramatically improve health outcomes and transform the delivery of healthcare. By generating evidence about the clinical utility of new tests and technology (such as PRS, ctDNA tests and newborn WGS), we can work towards developing new approaches to preventative healthcare.

As genomics research and innovation continues at pace, we are considering the potential applications in national policy and health and care services. The Office for Health Improvement and Disparities leads on prevention within the Department of Health and Social Care. The Office for Health Improvement and Disparities works closely with the UK National Screening Committee, OLS, NHS England and genomics delivery partners to ensure that the best research and evidence inform the development of policy and potential applications across health and care services to prevent or provide earlier detection of disease.

Our Genome UK strategy commitments are to:

- 11.** enable the NHS to move from a system that primarily detects and treats illnesses to one that utilises genomics to predict and prevent ill health
- 12.** continue to develop a public health and screening system that uses genomics to intensify screening and interventions in those at high risk
- 13.** establish a clear, evidence-based position on whether and how genomic sequencing should be implemented for newborns, and how that genomic data could inform their care later in life
- 14.** formulate a clear evidence-based position on whether and how PRS can be best utilised at scale in the health service
- 15.** explore how genomic testing can continue to be best used in reproductive medicine to support parents to make informed choices



### **Actions our genomics delivery partners have taken over the past 18 months**

- ▶ Genomics England has progressed discussions to inform the development of their newborn sequencing project via expert working groups to ensure successful delivery of the newborn genomes programme. Work has included engagement workshops and an online survey to understand views and inform the choice of conditions to screen for. This involved engaging with NHS clinician specialists who are advising how downstream treatment pathways would work, interviewing parents to understand experiences and attitudes, commissioning an evidence assessment and literature review on ethical issues, and running feasibility studies to assess the optimal approach to sampling and sequencing.
- ▶ Our Future Health has commenced recruitment, inviting volunteers to participate in the UK's largest ever health research programme via the NHS and other partners, and delivering appointments in partnerships with Boots and Acacium. To progress their PRS commitments, Our Future Health has awarded contracts for biological sample receipt and processing (UK Biocentre, Randox Laboratories Limited), genotype assay design (Illumina), genotyping service provider (Eurofins). They will collaborate with Genomics plc for imputation and PRS calculations.
- ▶ The UK National Screening Committee's evaluative roll-out of non-invasive prenatal testing for Down's syndrome, Edward's syndrome, and Patau syndrome as part of the fetal anomaly screening programme started in June 2021. Three NHS Genomic Laboratory Hubs carried out testing on behalf of the national network. This approach to prenatal testing could reduce the need for invasive tests which are associated with an elevated risk of miscarriage.

### **Actions our genomics delivery partners will take over the next 3 years**

- ▶ Genomics England will use £105 million of government funding in a landmark research programme, in partnership with the NHS, to study the effectiveness of using WGS to find and treat rare genetic diseases in newborns. The Newborn Genomes Programme will analyse the genetics of newborns to speed up diagnosis of treatable conditions, which could result in thousands of life-saving interventions. The research programme will also explore how babies' genomic data could be used for discovery research, focusing on developing new treatments and diagnostics for NHS patients. It will also explore the potential benefits and broader implications of storing a baby's genome over their lifetime. Genomics England is working with key stakeholders from a range of disciplines and with NHS England and further details on which trusts the programme will work with will be posted on Genomics England's website in early 2023 (Genome UK commitments 11, 12 and 13). So far, the programme has:
  - ▷ established the principles for the genes and variants to be included
  - ▷ identified a process for asking parents to consent for their newborn to be included in the study
  - ▷ researched the best way to take samples from newborns
  - ▷ worked with bioinformaticians to establish the strategy for analysis of the data
  - ▷ identified the pathways and systems needed to return results to parents and enable the right care and treatment pathway

- ▶ In partnership with Genomics plc, Our Future Health plans to leverage UK Biobank's whole genome sequence data to design and implement a bioinformatics pipeline and calculate polygenic and integrated risk scores. Our Future Health plans to genotype participants' blood samples using a single nucleotide polymorphism array which is optimised for the UK population and will include single nucleotide polymorphisms that contribute to PRS. The array will also be optimised to predict blood types and assess pharmacogenetic variants, offering the potential for participant healthcare insights, including the return of polygenic or integrated risk scores if they choose to receive them, in partnership with the NHS. Metrics will include participants recruited, samples genotyped and PRS calculated (Genome UK commitments 11 and 14).
- ▶ UK Biobank will seek to develop a secure and scalable imputation service within its cloud-based research analysis platform to enable detailed information contained within its 500,000 participant genomes be used to enrich data collected by other resources using lower cost genotyping assays. It is anticipated that Our Future Health, as well as other studies, will be able to use this service as part of an approved research project to enhance the data they collect to increase the impact of future research findings, and support continued innovation within genomic healthcare for the benefit of patients (Genome UK commitment 11).
- ▶ NHS England and Our Future Health will set up a joint group to support Our Future Health in the return of risk information to participants, generated by PRS, and understand the impact it may have on the NHS (Genome UK commitments 11 and 14).
- ▶ NHS England has developed a ground-breaking commercial partnership with GRAIL for the testing and use of their Galleri genomic test for cancer, aiming to accelerate the test into widespread usage as rapidly as the evidence allows. The agreement encompasses the trial stage (currently underway, 140,000 participants recruited) and an interim implementation phase, should early results from the trial hit specific benchmarks. As long as the research shows effectiveness, 500,000 tests will be rolled out in the financial years 2024/25 and 2025/26. Continued evaluation will be required as the test is used in more people in the NHS. The results will feed into the UK National Screening Committee's consideration for future screening programmes. NHS England is working closely with GRAIL to learn from the trial and ensure a smooth roll-out of the 500,000 tests (Genome UK commitments 11 and 12).
- ▶ NHS England has funded a transformation project through the NHS GMS Alliance, which will explore the implementation of ctDNA tests in the NHS, starting in stages 3 and 4 non-small cell lung cancer patients (Genome UK commitments 11 and 12).
- ▶ The UK National Screening Committee and NHS England will continue their 3-year evaluative roll-out of non-invasive prenatal testing for Down's syndrome, Edwards' syndrome and Patau's syndrome. The roll-out will be monitored to ensure any changes to the fetal anomaly screening programme pathway and screening processes can be recommended quickly and confidently by the UK National Screening Committee. Metrics for the programme have been published (Genome UK commitments 11, 12 and 15).
- ▶ The Genome UK prevention and early detection working group will work with the Office for Health Improvement and Disparities and genomics delivery partners within the group to ensure that they understand and can engage effectively with the relevant policy decision making processes to go from research through to policy and implementation (Genome UK commitments 11 and 12).

## Polygenic risk score (PRS)

A PRS is an estimate of an individual's genetic predisposition to a heritable trait, that is their risk of developing a disease. The information used to develop the score usually comes from genome-wide association studies which analyse large numbers of common genetic variants (single nucleotide polymorphisms) and their association with disease. A PRS typically comprises the sum of the effects of many single nucleotide polymorphisms (thousands or millions) across the genome into a single number, which is proportional to the individual's genetic predisposition for that trait. This can be combined with other information (such as age, sex or blood pressure) to create an integrated risk score. PRS and their application heavily depend on population-scale genomic biobanks such as UK Biobank, which has catalysed PRS research and studies of their clinical validity and utility.

Potential uses for PRS include:

- ▶ disease risk prediction – increasing the performance of disease risk models, or identifying individuals at significant genetic risk of disease but with few or no conventional risk factors
- ▶ screening – by predicting risk of developing disease and thereby enabling preventative interventions, or to allow risk stratification for existing screening programmes
- ▶ informing diagnosis and prognosis – improving diagnostic accuracy by predicting the subtype or severity of a disease
- ▶ management – helping to predict the response to a drug in order to guide treatment
- ▶ prompting risk reducing behaviours, such as increased exercise, weight loss, health system engagement and adherence to screening

The PHG Foundation has produced [a series of reports](#) which set out the potential applications of polygenic scores for risk prediction in different contexts, as well as outlining current gaps in the scientific evidence. So far there has been a lot of research on evaluating the performance of polygenic score models – that is, the standalone or incremental predictive value of a PRS on top of other established risk factors. To demonstrate the clinical utility of PRS, further implementation and translational research is needed into how PRS can be delivered within care pathways for specific conditions and target populations, as well as assessment of the outcomes.

## Polygenic risk score (PRS) – continued

Examples of programmes contributing to evidence around PRS are listed below.

- ▶ Our Future Health plan to calculate PRS for research and offer these, in a responsible manner, to participants who wish to receive them.
- ▶ UK Biobank genotyping data on 500,000 participants has enabled genomics research worldwide on an unprecedented scale that led to the concept of PRS being developed. Researchers found that about 5% of the UK Biobank population had a PRS which identified them as having a similar risk of developing heart disease to someone with familial hypercholesterolaemia. UK Biobank has since made PRS available for its 500,000 participants based on models developed by the wider research community for over 50 diseases areas, together with tools to evaluate PRS in a testing subset of the UK Biobank. When access to primary care data becomes available, researchers will be able to develop further PRS across a wider range of conditions and contribute to the growing evidence base.
- ▶ Genomics plc is a provider of PRS to the UK Biobank and Our Future Health programmes. In partnership with the NHS and GPs in the north of England, they recently ran a study called HEART which added PRS testing to the cardiovascular risk assessments – health checks – carried out in routine primary care in more than 800 participants. HEART evaluated the genomics integrated risk tool which combined a cardiovascular disease PRS with the currently used QRISK© method. GPs reported that the new tool was straightforward to incorporate into day-to-day practice, and that they and their patients found the test results offered helpful information. The results also found that 24% of all participants had clinically significant changes to their risk when genetics was added, leading GPs to report that they would change their management of 13% of study participants. The PRS models used were developed using UK Biobank data on 500,000 participants. These PRS models have been incorporated back into the UK Biobank resource together with evaluation tools to allow researchers worldwide to validate and develop further PRS models for an increasing range of diseases.
- ▶ Investigators at many universities and NIHR biomedical research centres are conducting research into multiple aspects of PRS, including the development of analytic methodologies, score optimisation, open translational resources and tools, as well as PRS implementation and delivery for particular diseases.
- ▶ There are international PRS efforts which focus on ethnic diversity, health equity and implementation, such as the National Institutes of Health PRIMED Consortium (USA), the National Institute of Health eMERGE Network (USA), the National Institute of Health All of Us Programme (USA), and the INTERVENE Consortium (EU Horizon 2020).

## Research and data

The UK is already a leader in genomic research. In Genome UK, we said that we would work to extend that lead by developing an ecosystem of world-leading, secure genomics datasets to drive research and support translation of research findings. These datasets will need to include more data from diverse ancestry groups, currently under-represented in genomic research. They should support studies that will improve our understanding of disease and help us develop new, more precise therapies. Over the next 3 years, we will move towards a future federated ecosystem by improved data standards and interoperability of genomic datasets for research use, including working through commercial principles for data access. The datasets will also be used to improve clinical trial recruitment so that patients will be able to benefit from improved access to genomically-informed trials. The action on our commitment to create a world-class offer on functional genomics, will further help to attract investment and grow the life sciences sector, in turn supporting economic growth.

Given the interdependency between genomic research and analysis of genomic data, the Genome UK cross-cutting theme for data has been integrated into the research pillar.

Our Genome UK strategy commitments are to:

- 16.** ensure that clinical genomic testing and genomics research contribute to powerful national data resources
- 17.** coordinate the UK's existing and future genomics ecosystem, enabling ground-breaking research at scale for the benefit of patients
- 18.** enable and empower genomics research, providing capabilities at a unique scale
- 19.** achieve greater diversity within our reference genomes, and future GWAS will reflect the UK's diverse populations
- 20.** incentivise the genomics research community to prioritise areas of high NHS unmet need
- 21.** support hypothesis-driven identification, recruitment, phenotyping, and biosampling of uniquely informative cohorts of patients
- 22.** develop consent and data standards that support innovation for the benefit of patients and the NHS, while maintaining trust in the safe, appropriate and responsible use of data
- 23.** work at a UK level to ensure there is equitable access to opportunities to participate in clinical trials informed by genomic data commitments

From the data cross-cutting theme:

- 24.** through the use of machine learning and AI, understand how genomically-informed healthcare and prevention could be improved and how these could be implemented in the NHS, embedding potentially lifesaving technologies quickly and efficiently in the NHS
- 25.** establish a clear set of standards for genomic and health data
- 26.** develop systems to enable federated access to data for research use to enable comparisons across multiple datasets

27. track the usage of our datasets and maintain an upward trajectory of both numbers and user experience
28. learn from the growing number of AI-based businesses in the UK on how to turn these applications into healthcare interventions

### **Actions our genomics delivery partners have taken over the past 18 months**

- ▶ UK Biobank completed WGS of all 500,000 participant samples at the end of 2021 as part of the world's largest sequencing project. Genome data for 200,000 genomes were released to approved researchers in November 2021, extending the genetic characterisation beyond existing cohort-wide exome and genotyping data, which will lead to exciting new insights. In collaboration with the funding industry parties, new data engineering methods have been developed to enable 'at scale' processing and curation of WGS data.
- ▶ UK Biobank has continued to develop its UK cloud-based research analysis platform in partnership with DNAnexus. The platform was made available to all researchers in September 2021 and is now being used by over 2,500 researchers. Leveraging technological methods developed with UK Biobank, DNAnexus has been selected to provide the underlying platform for the Our Future Health trusted research environments to provide access to its research data when these become available. UK Biobank continues to release new platform functionality to enable researchers to easily analyse the vast, multi-modal data held within UK Biobank's biomedical database.
- ▶ The NHS GMS Research Collaborative is a partnership between NHS England, Genomics England, the NIHR and the NHS GMS that aims to support genomic research on a national scale. The collaborative has [published](#) the process for submitting applications for the NHS GMS to support genomic research or pilot new technologies. They have to date received 10 proposals from across the NHS, academia and industry. Five proposals have also been received through the early feedback review service that enables researchers to use the expertise of individuals in the NHS GMS to inform their proposals. Early feedback requests are reviewed in an average of 18 working days. First draft capacity and capability statements have been developed by all NHS GMS Alliances to start to build a picture of the genomic research already underway across England. The collaborative has also formed a consent sub-group to develop a patient choice framework that makes consented non-WGS genomic data available for research.
- ▶ Following industry feedback about the importance of functional genomics capabilities for UK competitiveness, generating disease insights and improving drug discovery, the Medical Research Council, working with OLS, have engaged with academic and industry stakeholders to develop a functional genomics initiative with government funding secured.
- ▶ Genomics England has successfully concluded the first year of their diverse data project, delivering 5,000 samples from diverse communities. The programme has initiated engagement activities with representatives from potential future cohorts and other genomic institutions looking to develop initiatives in diversity.
- ▶ Genomics England has partnered with Lifebit to develop a new, cloud-based secure data environment which will provide improved functionality and usability for authorised researchers. This is being run alongside Genomics England's original research environment to ensure Genomics England can provide the right service to the broadest spectrum of use cases.

- ▶ NHS England's data for research and development programme was announced in March 2022. It includes up to £18 million, subject to government approval, to support data and research commitments in Genome UK to improve the use of genomics and related health data for research and innovation.
- ▶ The Department of Health and Social Care's [Data Saves Lives Strategy](#) was published in June 2022, which included the commitment to bring together genomics data and work with NHS England to ensure that genomic data generated through clinical care is fed back into patients' records.
- ▶ Development work has continued in the Global Alliance for Genetics and Health, including approval for consent clauses for large scale projects, using work from 14 countries. They are also involved in a series of outreach activities with major genomics centres to promote adoption of standards.

### **Actions our genomics delivery partners will take over the next 3 years**

- ▶ UK Biobank will be making its genome data available to industry partners and approved researchers, releasing the final genomes of their 500,000 total to approved researchers in the last quarter of 2023. This data will be available to analyse alongside over 15 years of follow-up health outcomes data, enabling researchers to further understand the genetic determinants of a wide range of diseases. Metabolomic and proteomics data are also being added to the UK Biobank resource, increasing characterisation of biological pathways and underlying disease mechanisms. UK Biobank will continue adding long-read sequencing and methylation assay data for the study of epigenetics to the resource. With £20 million funding from the Wellcome Trust, UK Biobank will also develop its research analysis platform, improving cloud-based platform functionality and going beyond genomic analyses to include more traditional analysis tools, such as imaging analytics interpretation of and health record data. This expansion of UK Biobank data availability will ensure that access to the resource, which is already one of the world's most genetically characterised and used research resources, becomes even more accessible and provides the necessary analytical tools to deepen our understanding of disease (Genome UK commitments 16 and 18).
- ▶ UK Biobank has secured funding for the world's largest longitudinal imaging dataset with £30 million committed by the Medical Research Council, Calico Life Sciences and Chan Zuckerberg Initiative. UK Biobank has already captured magnetic resonance imaging data from the brain, heart and abdomen, together with bone density and ultrasound scans of the carotid arteries, from over 60,000 participants. They aim to collect this data on up to 100,000 participants over the next years. This additional funding will allow repeat imaging to commence in the first quarter of 2023 on 60,000 participants, 2 to 7 years after their initial scan. When combined with the extensive phenotypic and deeply characterised genetic data already available in UK Biobank's database, repeat imaging data will advance understanding of the progression of a wide range of chronic diseases of mid-to-later life. It will also lead to improvements in diagnosis before symptoms even occur and enable the early interventions of potential therapies. UK Biobank is exploring linkages to existing digital pathology data collected within the NHS on its participants, with a pilot starting to link to digital histopathology slides for colorectal cancer patients in Leeds and Oxford (Genome UK commitments 16 and 18).

- ▶ In addition, UK Biobank has started a pilot with Wellcome Sanger Institute to undertake single-cell RNA sequencing on an initial 5,000 participants to enable research into functional genomics studies in individual cells. The pilot will conclude in early 2023 and, subject to funding, will be extended to 60,000 participants to undertake repeat imaging measures between now and 2028.
- ▶ Genomics England will receive £22.4 million government funding to carry out world-leading research, in collaboration with academic and commercial partners, to improve our understanding of genomic diversity and its impact on scientific, clinical and health system outcomes. This 3-year programme will increase the volume, depth and breadth of genomic data available from individuals belonging to ancestry groups that are currently under-represented in genomic research. This will be driven by tailored sequencing of 15,000 to 25,000 participants from diverse backgrounds by 2025, as well as community engagement work. Clinicians, analysts, researchers, patients and community groups will work together to develop new tools, processes and approaches for changing research, service-delivery practices, recruitment and care. The processes will be more equitable and the tools will be openly available to support international efforts, highlighting the UK's global leadership in genomics research. The diverse data programme aims to reduce health inequalities and improve patient outcomes within genomic medicine, improve genomics research with diverse populations, and earn trust of under-represented groups in genomics-informed personalised medicine (Genome UK commitments 16, 19 and 20).
- ▶ Our Future Health will aim to generate genotype array and genome-wide imputation data on up to 5 million participants. This will be combined with questionnaire and health-related linked data to create a research platform that enables discovery and translational research. Participants' consent includes the ability to re-contact them, enabling them to receive health-related insights be invited to additional studies based on disease risks. Our Future Health is aiming to recruit a cohort that reflects the UK population by age, ethnicity and socio-economic status, using census 2021/22 data as the comparison (Genome UK commitments 16, 19 and 21).
- ▶ The Medical Research Council has set up a new advisory group to support the scoping and design of an up to £25 million investment by UKRI-MRC in a 4-year functional genomics initiative, working across UKRI and other stakeholders. This initiative will respond to views from UK researchers and industry partners about the priorities for establishing a world-class offer on functional genomics, and build on existing infrastructure and UK research expertise. Open competition funding mechanisms are expected to launch in spring 2023 (Genome UK commitment 17, 18 and 21).
- ▶ NHS England will continue to work towards expanding the ability for researchers to access a range of genomics datasets through linkage of sources by scoping and testing Global Alliance for Genetics and Health interoperability modules with delivery partners. They will work with the NHS GMS on their interoperability programme to improve genomic test request processes and identify where processes could also improve research uses. They will also work with Genomics England on commercial principles around data access. This will be funded through the spending review allocation (Genome UK commitments 20 and 23).
- ▶ NHS England will continue to develop unrivalled at-scale data infrastructure (secure data environments) to deliver key research and development opportunities, making a variety of data types available in a streamlined secure and privacy-protected way. This includes work to scale up the NHS GMS' WGS capacity, enabling more clinical test sequences to be made available for research use (Genome UK commitments 16 and 18).



- ▶ NHS England is supporting faster, more effective and diverse data-enabled clinical trials by developing a service called Find, Recruit and Follow Up. The service will use data and digital tools to speed up the identification and recruitment of patients potentially eligible for specific clinical studies and enable follow-up. It will give a wider, more diverse cohort of the UK population the opportunity to take part in clinical research. Find, Recruit and Follow-up aims to address some of the challenges trialists face when they conduct studies in the UK, to reverse the decline in the number of studies taking place in the UK, and to enhance the quality of service. This service is one step towards creating a globally competitive, digitised, holistic and data-enabled clinical research process in the UK (Genome UK commitments 18, 21 and 23).
- ▶ The NHS will drive equity in access to clinical trials by aligning clinical trial targets with standard of care NHS testing. In appropriate circumstances this will involve partnering with clinical trial units and industry to identify eligible patients. This will require a mechanism to systematically horizon scan upcoming clinical trials to ensure the correct targets are added to the Test Directory, while also having the data sharing infrastructure in place to share genomic data safely where appropriate and with the necessary patient consent (Genome UK commitments 21 and 23).
- ▶ Over the next 2 years, NIHR BioResource will progress their long-term goal of creating a national infrastructure platform to enable the rapid recruitment to clinical trials and studies. Progress will be monitored through annual reporting, where metrics such as number of participants recruited and number of studies where NIHR BioResource has been used to support recall to studies are captured (Genome UK commitments 18, 19 and 21). They will use £40 million of NIHR funding to:
  - ▷ build and enhance phenotyping of disease cohorts, including rare diseases
  - ▷ increase inclusivity and diversity of existing cohorts
  - ▷ establish a Young People's BioResource
  - ▷ enhance and promote the offer to industry
  - ▷ develop new approaches to patient and public involvement
- ▶ The NHS GMS Research Collaborative will continue to use the NHS GMS infrastructure to facilitate a full spectrum of research and innovation, from discovery to translation, adoption and diffusion across the NHS. As part of the evolving NHS GMS Alliance infrastructure, the NHS will establish NHS genomic networks of excellence. These will bring together the NHS GMS, academia, universities, industry and other partners in networks to deliver genomic research from discovery to adoption and spread, in specific priority areas designated by NHS England and aligned to NHS priorities (Genome UK commitments 17 and 19).

- ▶ In partnership with Genomics England, patients and clinicians, NHS England have developed a national patient choice framework that supports clinicians, regardless of clinical specialty, to discuss the implications and impact of having WGS and whether a patient would consent to their genomic data being accessible for research via the National Genomic Research Library. To date, of patients undergoing WGS in the NHS who have been offered the opportunity to participate in research, approximately 93% of patients have given their consent. NHS England is working with partners to put in place mechanisms for enabling consent and collation of NHS genomic sequencing data for research and innovation purposes at a national and regional level (Genome UK commitment 22).
- ▶ As set out in the '[Life sciences competitiveness indicators 2022: life science ecosystem](#)', health data facilitates medical research and diagnostics. This can enable the development of treatments and earlier detection of disease. A rich supply of health data can allow for analysis of key health indicators, including genomics, to diagnose disease earlier when it is easier and less expensive to treat. High-quality data and associated architecture can bring together datasets to allow more detailed research and development of artificial intelligence and health technologies. There are currently no metrics available for the information environment. OLS is therefore considering how the UK data environment can be measured against other countries (Genome UK commitment 27).
- ▶ NIHR, the Medical Research Council and the Wellcome Trust have provided funding to the Global Alliance for Genetics and Health to develop standards and policies for sharing genomic and related health data. The Global Alliance for Genetics and Health aims to develop secure technical standards and frameworks to promote responsible use of genomic data for the benefit of human health, and drive uptake of standards through effective communications, dissemination and engagement. In the UK, Global Alliance for Genetics and Health standards are already being actively deployed within Genomics England, enabling better communication with Genomic Laboratory Hubs, the General Medical Council, and the broad NHS (Genome UK commitments 22 and 25).

## Case study



### Genes and Health

Genes and Health is a long-term, population health resource of adults, combining genetic data and lifetime multisource NHS health record data (primary care, hospital, and national NHS Digital) with the ability to invite volunteers to return with consent for more detailed research studies.

Genes and Health is researching British-Bangladeshi and British-Pakistani ethnic minority groups who have marked health inequalities (such as the highest rates of type 2 diabetes and early heart disease in the UK) and who are poorly represented in other large genetic research studies to date. Without such resources, modern genomic medicine and precision medicine (such as disease risk prediction) might not benefit communities with the greatest need.

The resource is open to international scientific researchers. Currently, over 85 groups of academic and industry researchers working across multiple disease and basic science fields are approved to analyse Genes and Health data via a UK cloud-based secure data environments. Genetic data includes chip genotyping and exome sequencing on all volunteers.

The first East London Genes and Health volunteer took part in 2015, with Bradford Genes and Health opening in 2019 and Manchester Genes and Health in 2022. There are now over 53,000 Genes and Health volunteers, and a target of 100,000 by 2024. Genes and Health is embedded in the local communities it is studying, with a wide-reaching and authentic programme of engagement activities. Their community advisory group works closely with the Genes and Health Executive to prioritise research topics and build acceptance and long-term support.

Recall studies to date include:

- ▶ 32 volunteers at very high risk of heart attack returned a diagnosis of low density lipoprotein receptor familial hypercholesterolemia with appropriate preventative treatment (none of whom were previously aware of their genetic diagnosis and risk level)
- ▶ laboratory studies on an individual lacking the HAO1 protein provided key safety information and biological insights for a new drug, lumasiran
- ▶ over 1000 volunteers identified for a blood ‘cell atlas’ sequencing project at the Wellcome Sanger Institute

## Case study



### NHS DigiTrials

NHS England has funded [NHS DigiTrials](#) (a delivery partner of Find, Recruit and Follow Up) which offers data services to support high-priority, large-scale clinical trials. DigiTrials reduces the time, effort and cost of developing new drugs, treatments and services, bringing benefits to patients, the public and the NHS. NHS DigiTrials' services can be used to accelerate the recruitment of diverse trial participants and increase the number of people identified as potentially eligible to participate in trials.

The NHS-Galleri trial is studying the clinical and economic performance of the Galleri™ test using healthy NHS volunteers. The cohort needed for the study was challenging to reach via normal clinical settings and had to align with specific demographic and cancer risk factors. NHS DigiTrials has supported recruitment by identifying eligible participants using routinely collected NHS Digital data. By July 2022, the service had recruited 140,000 volunteers across 8 areas in England in just 10 months, making it one of the fastest recruited large-scale randomised trials.

NHS DigiTrials is also supporting recruitment to the Our Future Health research programme. Millions of adults from all backgrounds will be invited to take part in the programme by providing a blood sample, information about their health and lifestyle, and their consent to link their NHS records. This will be used to create a detailed picture that represents the whole of the UK, helping researchers to discover more effective ways to predict, detect and treat common diseases. NHS DigiTrials will support recruitment by using data to identify people who are eligible and inviting them to join. Letters are sent directly to eligible participants to see if they want to take part in the programme. This means that no patient data leaves NHS Digital or is shared with the research programme.

## Cross-cutting themes

### Engagement and dialogue with the public, patients and our healthcare workforce

As we move forward in implementing our vision for genomic healthcare, it will be essential to bring patients and the public with us through continued engagement activities. Patient and public engagement is built into the governance of the major organisations that deliver Genome UK, such as Genomics England, NHS England and Our Future Health. We are now considering how patient engagement should be approached by Genome UK's governance structures to ensure that their voice is embedded into our decision making.

Our Genome UK strategy commitments are to:

29. ensure that patients, the public and the NHS workforce have an increased awareness and understanding of the potential benefits of genomic healthcare by increasing its visibility and committing to open, honest engagement about what is involved
30. set out clearly how patient data can be used to advance research and inform the public about research that has successfully used their data to improve diagnosis, understanding or treatment of patients in the UK
31. ensure that appropriate measures are in place to protect patient privacy and confidentiality, so that patient data are used in ways that are acceptable to the public

The following commitment from the ethics theme is also relevant here:

42. keep an open dialogue and continue to openly engage with relevant patient and participant groups, continuing to involve the public, building on the engagement through the 100,000 Genomes Project

### Actions our genomics delivery partners have taken over the past 18 months

- ▶ NHS England's NHS GMS people and communities forum has held regular meetings, with topics such as the NHS Genomic Strategy, Test Directory, clinical genomics service specification, WGS and consent, and the NHS GMS Research Collaborative discussed.
- ▶ Genomics England has continued to regularly engage their participant panel in addition to engagement around the newborn project. Engagement for the newborn project has included:
  - ▷ running an online survey that received over 600 responses
  - ▷ holding workshops with members of the public, people living with rare genetic conditions, and healthcare professionals
  - ▷ running a series of sessions with genetic counsellors and regional meetings with clinical and other specialists to explain the draft principles
- ▶ In July 2021, Genomics England published the results of a public dialogue on the use of WGS in newborn screening, finding that members of the public were broadly supportive as long as the right safeguards and resources are in place.

- ▶ UK Biobank has completed a consultation with the UK's public engagement charity, Involve, to identify opportunities for greater participant involvement and engagement as part of study governance and future enhancements. In addition, its ethical advisory committee has started to bring together a focus group to assess participant views on extending linkages to health-related records and, specifically, access to participant tissue samples that may have been collected within the NHS.
- ▶ Our Future Health has held regular meetings of their public advisory board, feeding into aspects of the programme including consent revisions, trusted research environment plans and pilot evaluation. Public representatives have also joined other Our Future Health advisory boards, including their ethics advisory board and technology advisory board.

### **Actions our genomics delivery partners will take over the next 3 years**

- ▶ The NHS GMS will continue to drive the proactive involvement of patients and the public from our diverse communities, nationally through the NHS GMS people and communities forum and regionally throughout the NHS GMS infrastructure (Genome UK commitments 29 and 42).
- ▶ NIHR BioResource will increase patient and public involvement in the review of applications, support participant recruitment through their participant portals, work with under-served communities and support patients to develop their research ideas into research projects. Each BioResource cohort uses innovative ways to recruit participants. For example, the Young People's BioResource has developed a young ambassador programme to help a group of young people promote the aims of the Young People's BioResource to their peers and to support recruitment. Recruitment to this programme started in September 2022 (Genome UK commitments 29 and 42).
- ▶ OLS is leading work on how best to engage patients in its Genome UK implementation co-ordination group and associated working groups (Genome UK commitment 42).
- ▶ UK Biobank will further expand its patient and public involvement activities. Following a review it commissioned with the UK's public participation charity, Involve, it is exploring additional ways it can inform and involve its 500,000 participants as part of the ongoing study and future enhancements (Genome UK commitments 29 and 42).
- ▶ Our Future Health will continue to grow their public and participant involvement by having representatives in their wider governance structures. They will co-develop and co-design policies and procedures with their public and participant representatives, as well as involving members of the public in regular user testing of participant-facing materials. Our Future Health will engage with communities and partners to increase awareness of the programme in order to maximise participation, particularly from minority populations that have historically been under-represented in large-scale, population-based studies (Genome UK commitments 29 and 42).

- ▶ The national data advisory group has been established and is now meeting routinely. Membership draws from across expert external health and care stakeholders, as well as patients and regional system representatives. The responsibilities of the group include:
  - ▷ testing approaches and thinking for national programmes and policy areas, including what topics should be engaged on and how
  - ▷ providing advice on national strategic products, including on the engagement standard for public engagement
  - ▷ considering how national strategic work can support local and regional teams on data issues
  - ▷ national strategic communications advice, including tone and focus
  - ▷ advising on national strategic stakeholder engagement and co-design work (Genome UK commitments 30 and 31)

### **Workforce development and engagement with genomics through training, education and new standards of care**

The genomics workforce spans both the health service and industry. It includes laboratory-based staff such as clinical scientists, genomic technologists and bioinformaticians, specialist clinical staff such as clinical geneticists and genetic counsellors, and members of the mainstream workforce who encounter genomics in their role, such as doctors (including general practitioners), pharmacists, nurses and midwives. Each of these professions play a vital role in the genomic healthcare ecosystem, and continued proactive efforts are required to ensure that they have the support and resources needed to deliver genomic advances to patients now and in the long term.

NHS England, working in partnership with Health Education England (HEE) and the Department of Health and Social Care, are currently developing a long-term workforce plan for the NHS, as commissioned by the government earlier this year.

Our Genome UK strategy commitments are to:

- 32.** ensure that all new graduating doctors, nurses, midwives, pharmacists, allied health professionals, dental and relevant nonclinical staff have a level of awareness and knowledge of genomics that is relevant to their role
- 33.** ensure that the healthcare science workforce continues to have advanced genomic training and education within their programmes
- 34.** put in place continuing professional development (CPD) programmes to ensure all relevant staff maintain an up-to-date and role-appropriate understanding of genomics
- 35.** use workforce modelling data to inform investment decisions for training numbers across all professions and support workforce growth to meet the needs of the NHS GMS, particularly in specialist scientific and medical workforce areas
- 36.** establish and invest in training pipelines for in-demand occupations such as bioinformatics to build capacity within the health service and the wider sector

**37.** redevelop clinical pathways and standards of care to that fully incorporate the latest genomic testing and results.

**38.** support the NHS workforce by providing simple, practical, informatics solutions for training, genomic analysis and decision-support

### **Actions our genomics delivery partners have taken over the past 18 months**

- ▶ NHS England and HEE undertook a joint workforce data capture exercise for the NHS Genomic Laboratory Hub workforce between July and September 2021. This data will be used to inform supply and demand modelling.
- ▶ HEE surveys aimed at the pharmacy and the nursing and midwifery workforces were launched. The findings will help HEE to understand the levels of interaction with genomics in practice, and gaps in knowledge can then be addressed through the Genomic Education Programme's strategic approach to workforce development.
- ▶ HEE has developed their clinical pathway initiative in collaboration with NHS England and the Academy of Medical Royal Colleges. The clinical pathway initiative outlines a stepwise approach to multi-professional clinical pathways, identifying the workforce associated with each touchpoint along the pathway and the education and training interventions required where there are gaps in knowledge or competency. The clinical pathway initiative provides a platform for sharing workforce education and training needs across different clinical pathways to support the workforce and avoid duplication across the system.
- ▶ A joint HEE and NHS England spending review bid resulted in funding for additional genomics-related scientist training programme and higher specialist scientist training places, an increase in practice educators, and the establishment of a Genomics Training Academy.
- ▶ An NHS England and HEE pharmacy genomics workforce group has been set up to provide a forum for national collaboration and co-ordination of pharmacy workforce planning and education activity related to genomics. A pharmacy genomics roundtable, hosted by HEE's Genomic Education Programme and NHS England, was held in November 2022.

### **Actions our genomics delivery partners will take over the next 3 years**

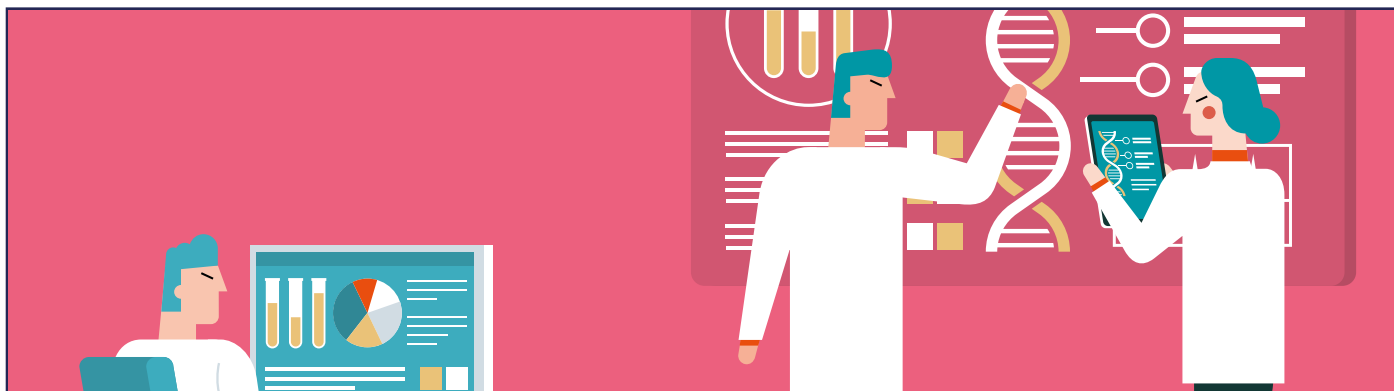
- ▶ To ensure that genomics is represented in the undergraduate curricula of the mainstream workforce, HEE will:
  - ▷ scope the existing undergraduate curricula for medicine, nursing, midwifery, and dentistry
  - ▷ work with higher education institutions to update curricula to integrate genomic medicine
  - ▷ provide 'off the shelf' packages to support the delivery of genomic education and training in the undergraduate setting (Genome UK commitment 32)



- ▶ To ensure that the specialist genomic workforce have access to the continuing professional development required for their roles, HEE and NHS England are establishing a Genomic Training Academy. Over the next 3 years this will involve developing education and training resources mapped to profession-specific curricula. Metrics will include:
  - ▷ establishment of the Genomic Training Academy and infrastructure
  - ▷ numbers of modules and teaching events developed and delivered
  - ▷ numbers of workforce who have benefitted from Genomic Training Academy training
  - ▷ evaluation of Genomic Training Academy and resources (Genome UK commitments 33 and 34)
- ▶ HEE will utilise NHS England and Genomic Education Programme workforce modelling data by increasing scientist training programme numbers across the specialist genomics workforce (laboratory and clinical) and developing and delivering new models of genomic education and training provision to ensure the offering is relevant to different specialties and professions. Increase in scientist training programme numbers will be funded through Spending Review 2021 allocation. HEE will also investigate how to retain bioinformaticians through new models of working. Metrics will include:
  - ▷ numbers of clinical scientists in post
  - ▷ numbers of WGS cases going through the laboratories
  - ▷ retention of current staff (Genome UK commitments 33, 34, 35 and 36)
- ▶ HEE will support clinicians to use the Test Directory by developing 2 massive open online courses aligned to the rare disease and cancer genomic pathways and continuing the development of their GeNotes resource. GeNotes consists of 2 tiers. Tier 1 is mapped to the Test Directory and supports the clinician to choose the right genomic test for the right patient at the right time and navigate the Test Directory and its supporting resources. Tier 2 is 'the knowledge hub', providing an extended learning opportunity for clinicians engaging with the resource and content. Metrics will include the number of people accessing the course, as well as evaluation of the course (Genome UK commitments 37 and 38).
- ▶ HEE will actively work to bridge the clinical-research gap through monthly blogs where they discuss a particular aspect of genomic research and its clinical impact, through the establishment of a new 'expert webinar' series and through the development, funding and collaborative delivery of the masters in genomic medicine framework (Genome UK commitment 34).
- ▶ HEE will also be horizon scanning to determine where new research findings may impact on clinical practice and using this to inform workforce modelling and clinical pathways. HEE will collaborate with Royal Colleges and the Academy of Medical Royal Colleges to ensure that new advances are prospectively represented in curricula (Genome UK commitments 35 and 37).
- ▶ The NIHR clinical research network's medical directorate is committed to helping ensure the NHS workforce is competent in delivering genomic research. They are working with specialties and NIHR Learn, as well as with the Academy to develop training curricula and materials for busy clinicians (Genome UK commitments 33 and 34).

- ▶ OLS and industry continue to explore how the skills value chain approach could support the adoption of emerging skills in the sector. This work will encompass current areas of shortage, such as bioinformatics, data analytics and computational biology (Genome UK commitments 35 and 36).
- ▶ A strategy outlining the approach to supporting educational and training needs for the pharmacy workforce will be published in early 2023 (Genome UK commitment 34 and 35).
- ▶ The national nursing and midwifery genomic transformation programme, led by the NHS England genomics unit, has been commissioned to provide a 2-year programme of activity from 2022 to 2024. The programme will engage with hundreds of nurses and midwives to support the development of their knowledge and skills in genomics, building their confidence and capability to lead, deliver and co-ordinate genomic practice in everyday care. The programme will also support nursing and midwifery leaders to define exemplar genomic pathways and accelerate the adoption of standardised practice at appropriate clinical touchpoints to increase equity of access or reduce unwarranted variation (Genome UK commitments 34 and 37).

## Case study



### Apprenticeships

The BiIndustry Association's 2022 [Genomics Nation report](#) included a 'spotlight on skills' which quoted that 70% of UK genomics small and medium-sized enterprises (SMEs) relying on the full range of skilled professionals say that it is particularly difficult to recruit for computational or data skills. Apprenticeships offer development of highly sought-after informatics skills in existing biotech talent within the industry context.

Cranfield University has been offering a bioinformatics masters-level apprenticeship since 2019 and postgraduate training in bioinformatics since 2002. The course features a strong focus on genomics and genetics as well as computational skills, providing life science companies with the opportunity to address these skills gaps within their organisations.

Freeline – a clinical-stage biotechnology SME – identified a high achieving research assistant with an aptitude for computational sciences who wanted an opportunity to develop their skillset. The employee was keen to bridge the gap between biology and computer skills, with the aim of being able to aid the Freeline team in discovering and developing new ways to deliver gene therapies for patients. The employee and their line manager therefore actively sought out Cranfield's apprenticeship programme, with the employee joining as a part-time master's student.

Freeline have found that the bioinformatics apprenticeship provides a structured forum for the employee to develop their knowledge, technical and computational skills. Employees and their line managers meet with the course organisers to discuss progress every 3 months. The employer also works with the course organisers to develop a bioinformatics research project that is relevant to the company and forms part of the evaluation of the course. An advantage of the course is that the exercises carried out by the apprentice are grounded in real-world data, with Freeline's employee working to develop tools that can be practically used in the company's pipeline.

## Case study



### **BioIndustry Association's Manufacturing Advisory Committee Leadership Programme**

The BIA Manufacturing Advisory Committee Leadership Programme supports the development and training of managers in the biopharmaceutical and cell and gene therapy industries through cross-sector learning and peer networks, helping deliver future leaders.

Two key aims of this initiative are:

- ▶ to promote cross-sector learning by offering an overview of the work of other companies across biopharma, vaccines, and cell and gene therapies by seeing them in action
- ▶ to develop a network with peers to share best practice and develop relationships to encourage possible future collaborations

The pilot programme was launched in January 2017 and completed in January 2019. An alumni group was set up afterwards to support networking. There are currently 92 participants from 36 member companies benefitting from the programme.

### **Supporting industrial growth in the UK**

The UK has a thriving genomics industry and, as set out in Genome UK, we are committed to making the UK the best location globally to start and scale new genomics healthcare companies and innovations. To deliver this, it is essential that engagement with companies across the sector is embedded in genomic healthcare across diagnosis and personalised medicine, prevention and early detection, and research and data and that the delivery partners for Genome UK have a strong relationship with sector leaders to continue to support growth.

We have held workshops with representatives of the genomics industry to identify top priorities, resulting in clear actions for delivery partners, and an increased understanding of how best to support our world-leading genomics industry. We will continue to take this approach in future years.

Our Genome UK strategy commitments are to:

39. develop integrated data resources, biosampling capabilities, and collaborative academic and clinical expertise that will make the UK the most attractive location globally for genomic healthcare start-ups
40. help to increase life science industry research and development spend in the UK by identifying new opportunities for innovative and cutting-edge industry partnerships
41. work to improve the availability of capital, including through the Life Sciences Investment Programme, which will deliver around £600 million of investment – both public and private – to be deployed with a significant focus on UK Life Sciences companies over the next 10 to 15 years

### **Actions our genomics delivery partners will be take over the next 3 years**

- ▶ As set out in the [shared commitments for implementation](#) published in March 2022, OLS and its industry delivery partners, including those based in the devolved governments, committed to holding a joint workshop in partnership with the trade associations, the Association of the British Pharmaceutical Industry and the BioIndustry Association. Two workshops have already taken place during 2022, with further workshops planned to gather industry feedback on Genome UK implementation and better understand industry priorities.
- ▶ OLS will continue to evaluate their [bioscience and health technology sector statistics](#), which publishes information on the shape and size of the genomics sector in the UK to ensure it is meeting user needs and evolving alongside the changing genomics landscape. OLS will work with users and delivery partners to collect feedback and ensure the statistics provide the most accurate reflection of the sector and continue to measure how the sector is changing.
- ▶ OLS, through the [bioscience and health technology sector statistics series](#) and the BioIndustry Association's Genomics Nation reports, have published statistics monitoring the activity of genomics companies in the UK. There are some differences in the definitions of genomics companies in these published statistics. The BioIndustry Association and OLS will work together to understand these differences and establish a consistent approach to measuring and reporting on the size, makeup and growth of the sector to inform policy development. More information on what companies are included in the OLS statistics can be found in the accompanying 2021 [user guide](#) and within the subsectors chapter of the BioIndustry Association's [Genomics Nation Report](#).
- ▶ The Life Sciences Scale-Up Taskforce, co-ordinated by OLS and supported by the BioIndustry Association, produced tangible actions the government and industry could take forward to increase the availability of capital for life sciences, including genomics companies. This included exploring mechanisms that bring together institutional investors and specialist venture capital firms to attract more private investment towards specialist funds supporting innovation, to strengthen the UK's investment ecosystem, and to help growing companies scale in the UK (Genome UK commitment 41).

- ▶ Delivery partners for Genome UK will continue to identify new industry partnership opportunities. This will include aiming for equitable opportunities for SMEs, to enhance UK attractiveness and continue to support growth. We will assess the process for SME access to data resources, bio sampling capabilities and expertise. We will also take action to simplify the application process for small businesses which want to supply to government and to increase visibility of subcontracting opportunities (Genome UK commitment 40).

## Case study



### Genomics England working with SMEs

Genomics England has a mandate from our participants, expressed clearly by the participant panel and Access Review Committee, to provide access to the National Genomic Research Library for the biopharma industry on the basis of fair economic return. For this reason, Genomics England charges an access fee for our commercial partners, and 8 of the top 10 pharmaceutical companies in the world pay to access our data. We also recognise the huge contribution to research and development made by the SME industry, including start-up companies. For this reason, SMEs are charged one sixth of the rate of large biopharma partners for access. Furthermore, Genomics England have partnered with 3 accelerators that focus on early-stage start-ups with a specific genomics focus in cancer and rare disease. These accelerators provide an effective filter for identifying some of the best start-ups. For these start-up and pre-revenue companies, Genomics England provides zero-rated access for the first year, which can be extended at Genomics England's discretion beyond 1 year.

For example, Nostos Genomics is an artificial intelligence-driven start-up that is looking to use sequencing data to cut rare disease diagnosis time by 99% by automating the identification of disease-causing mutations in patients' DNA. This could cut costs for labs, free up resources and, crucially, improve patient care. Nostos is using Genomics England's rare disease dataset as a training and validation set. The company is at an early stage of clinical validation, but the results look promising. As a result, Genomics England has extended zero-rated access for a second year with Nostos Genomics.

## **Maintaining trust through strong ethical frameworks**

In implementing genomic healthcare, we want to harness the power of genomic and genetic information combined with other health data to be able to provide more timely, improved diagnosis and offer better, equitable and more personalised treatments and access to clinical trials. To enable these advances, it is important that the public and patients can be reassured that ethical questions including those regarding consent, confidentiality and the handling of genomic data in research have been considered in a comprehensive way, with public and patient participation, and that these questions are addressed with robust data governance and secure data protocols.

Together with the devolved governments, we agreed to work together on embedding ethical considerations in both genomic healthcare policy development and programme planning and implementation. To achieve this, we said we would hold a series of workshops later in 2022, in collaboration with the Nuffield Council on Bioethics.

Our Genome UK strategy commitments are to:

- 42.** keep an open dialogue and will continue to openly engage with relevant patient and participant groups, continuing to involve the public and building on the engagement through the 100,000 Genomes Project
- 43.** establish a gold standard UK model for how to apply strong and consistent ethical and regulatory standards. We will share these standards and expertise globally and help partners across the world develop and implement their own frameworks
- 44.** ensure that our regulatory and ethical frameworks support rapid healthcare innovation, whilst reflecting legal frameworks and retaining public and professional trust. We will keep under review the balance between regulation and innovation

### **Actions our genomics delivery partners have taken over the past 18 months**

- ▶ The Nuffield Council has led work on gathering case studies and convening stakeholder workshops, which took place in December 2022. The workshops explored the feasibility and development of a UK model for how to apply strong and consistent ethical standards in genomic healthcare and research.

### **Actions our genomics delivery partners will take over the next 3 years**

- ▶ In July 2022, the Nuffield Council on Bioethics issued a call for case studies from the past 10 years describing how organisations have dealt with ethical issues they have encountered during their work on genomics initiatives. The case studies cover examples from across the NHS, government, academia, and the charity and private sectors. They were analysed for common themes and examples of best practice, and were discussed at the UK-wide workshops in December 2022. A report of the outcomes and an illustrative selection of the case studies will be published on the Nuffield Council on Bioethics website in early 2023. This work is aimed at scoping the feasibility and development of a UK model for ethical standards in genomics initiatives and will lead to a better understanding of ethical issues encountered in genomics research and how to overcome them (Genome UK commitments 43 and 44).

- ▶ Genomics England has expanded its ethics team and is developing its approach to embedding ethics more substantively in its research activities. This includes refreshing the Access Review Committee for the National Genomic Research Library, co-producing policies on issues such as recontact with input from the participant panel, and streamlining processes for internal ethics advice in collaboration with the legal and information governance teams.
- ▶ The Newborn Genomes Programme at Genomics England is embedding ethics operationally in the study to ensure an ethical study design, implementation and evaluation. The programme has an ongoing independent newborn ethics working group to help identify ethical considerations and support the development of new policies. The programme is also commissioning research including a literature review on the ethics of sequencing newborns and the regulatory and governance aspects of the lifetime genome to inform future thinking and delivery of the programme. Other supporting public dialogue will be undertaken on specific areas – for example, to explore public views on the acceptability and scope of the discovery research potential for this cohort.







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