- Department for Business, Energy & Industrial Strategy (https://www.gov.uk/government/organisations/department-for-business-energy-and-industrial-strategy)
- Department of Health (Northern Ireland) (https://www.gov.uk/government/organisations/department-of-health-northern-ireland)
- Department of Health & Social Care (https://www.gov.uk/government/organisations/department-of-health-and-social-care)
- Office for Life Sciences (https://www.gov.uk/government/organisations/office-for-life-sciences)
- Scottish Government (https://www.gov.uk/government/organisations/the-scottish-government)

- Welsh Government (https://www.gov.uk/government/organisations/welsh-government)

Policy paper

Genome UK: shared commitments for UK-wide implementation 2022 to 2025

Published 18 March 2022

Contents

This publication is available at https://www.gov.uk/government/publications/genome-uk-shared-commitments-for-uk-wide-implementation-2022-to-2025/genome-uk-shared-commitments-for-uk-wide-implementation-2022-to-2025

Ministerial foreword

The UK is a global leader in genetics and genomics. This has never been more evident than in the last 2 years, where collectively we have led the world in virus and human genome sequencing to counter the COVID threat and added 500,000 whole genome sequences to the UK Biobank research dataset.

In 2020 we published our overarching Genome UK – the future of healthcare (https://www.gov.uk/government/publications/genome-uk-the-future-of-healthcare) strategy, which set out our vision and clear aspirations for how we will transform genomic healthcare over the next 10 years. 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 demonstrating successful delivery. To achieve this progress, we have set out here a series of shared commitments for UK-wide implementation. We are committed to working together along with our delivery partners across the UK to implement these commitments, and, in doing so, realise the potential of genomic healthcare for the benefit of patients across the UK and around the world.

In developing these shared commitments, we have engaged in open dialogue and collaboration across the UK, recognising the differences in our respective healthcare systems and structures.

We believe that these shared commitments will help to ensure better coordination of our joint ambitions for genomics research and healthcare so that these can flourish in each of our nations and across the UK. Through better UK-wide coordination and collaboration, we will further strengthen our ability to share expertise and establish new collaborations and partnerships with others worldwide, securing our global leadership in genomics and the wider life sciences and ensuring we remain an attractive location for research and development investment. These shared commitments present a clear statement of our resolve to work together to deliver better health outcomes across the UK.

Background

In September 2020, the UK government published Genome UK – the future of healthcare (https://www.gov.uk/government/publications/genome-uk-the-future-of-healthcare), setting out the government's 10-year strategy to create the most advanced genomic healthcare system in the world, delivering better health outcomes at lower cost. The strategy also describes a vision for the UK to be the best location globally to conduct genomics research and grow new genomics healthcare companies, with a goal to increase private sector investment.

We want to ensure that patients across the UK can benefit fully from genomic healthcare, through a more preventative approach, faster diagnosis, and personalised and better treatment leading to better long-term outcomes. Researchers and industry will be supported in their research and its applications and incentivised to secure the UK's position at the forefront of genomic research in the world.

In May 2021, the UK government published its 2021 to 2022 implementation plan (https://www.gov.uk/government/publications/genome-uk-2021-to-2022-implementation-plan) for Genome UK, setting out priority actions for the financial year 2021 to 2022 in England, with contributions from the Scottish and Welsh Governments outlining their approach to implementation.

Genomics is a fast-moving field and we have therefore adopted a phased approach to research and implementation which will allow us to review our commitments and take action to reflect emerging science and latest research findings. The 2021 spending review, which set departmental budgets and devolved government allocations to 2024 to 2025, provides an important and timely opportunity to collectively agree high-level commitments with which we will progress implementation of the Genome

UK vision over the next 3 years. 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 demonstrating successful delivery.

There are many areas where UK-wide collaboration in genomics has already been successful, the SARS-CoV-2 genome sequencing in response to the COVID-19 pandemic provided an excellent example of this. In the coming years, UK-wide coordination will continue to provide significant opportunities to enhance benefits for patients, such as joint genomic technology evaluation and better integration of genomic and health data in secure trusted research environments.

We remain committed to delivering genomic healthcare across the UK, while recognising the devolved nature of healthcare policy and the resulting different approaches to the development of genomics in healthcare. In this context, our shared commitments form part of our second phase of Genome UK implementation, setting out joint, UK-wide, high-level commitments for the period 2022 to 2025. Recognising the devolved responsibilities, the shared commitments will be followed by 4 separate implementation plans, with the UK government and the devolved governments each aiming to publish these by the end of 2022. The separate implementation plans will reference the shared commitments, incorporating and building on them, in addition to setting out more detailed commitments for each government.

Commitment principles, oversight and governance

In taking these commitments forward, we will be guided by the 8 shared principles stated in Genome UK and in particular the following principle, which will underpin our approach to working together:

We will work together across the UK to realise the potential of genomics for the benefit of patients and ensure that the genomics services thrive in each nation. We will engage in open dialogue and collaboration, recognising that health is devolved and there are differences in NHS structures and systems.

The shared commitments have been developed in collaboration with the genomics community and our delivery partners. High-level coordination and delivery progress will be considered by the Genome UK Implementation Coordination Group which is led by the Office for Life Sciences and has UK-wide representation. This arrangement will allow the UK government and the devolved governments to continue with, or put in place, their own reporting and governance arrangements.

The National Genomics Board is chaired by the Minister for Technology, Innovation and Life Sciences in the Department of Health and Social Care (DHSC) and brings together senior decision makers and representatives from across the genomics sector, including senior officials from the devolved governments. The purpose of the board is to provide strategic oversight and to work collaboratively across the UK to harness the benefits of genomic healthcare – ultimately helping to ensure delivery of the vision set out in Genome UK.

As part of these shared commitments, we agree that UK government and devolved government ministers will engage regularly on the outcome of National Genomics Board discussions.

Finally, the UK genomics healthcare policy landscape is vibrant and complex, with a wide range of diverse organisations either delivering or overseeing clinical services and policy programmes. To progress the commitments in this agreement, we will seek to minimise duplication of effort and resource by sharing information about existing processes, groups and structures and, with mutual agreement, utilise these when appropriate.

The shared commitments

The following commitments are set out across the 3 pillars of Genome UK:

- diagnosis and personalised medicine
- prevention and early detection
- research

And the 5 cross-cutting themes covering:

- ethics and maintaining trust
- engagement and dialogue with patients and the public
- data
- workforce development
- industry

Diagnosis and personalised medicine

Genome UK vision: to help people live longer, healthier lives by using genomic technologies to identify the genetic causes for disease, to detect cancers earlier and provide personalised treatments to illnesses.

Genomic technologies are already revolutionising the way in which patients are diagnosed and treated across the UK and this is set to accelerate rapidly in the years ahead.

At the same time, the devolution of healthcare and clinical service commissioning means that there are differences in how genomic healthcare has so far been implemented across the UK. However, we have a wealth of experience and leadership in diverse areas of genomics that can be shared across the UK to drive improvements in patient care. For example:

- the NHS in England, supported by Genomics England, is rolling out the world's first whole genome sequencing service for adults and children with certain cancers or indications suggesting an undiagnosed rare disease
- in Northern Ireland, collaboration between academia and Health and Social Care Northern Ireland has led to the creation of an integrated molecular diagnostic service to provide comprehensive genomic profiling to patients with solid tumours and haematological malignancies as well as those with inherited genetic disorders. Further work is ongoing to connect genetic and genomic information with electronic health records to allow genomic findings to be translated into improved patient management decisions and outcomes
- in 2020, Wales launched the UK's first rapid whole genome sequencing service for seriously ill children (WINGS – the Welsh Infants' and Children's Genome Service), the first pharmacogenomics service in the UK, routinely screening all cancer patients eligible for treatment with fluoropyrimidine-based chemotherapy to identify risk of severe side-effects, and rapidly established SARS-CoV-2 sequencing service to support the pandemic
- in Scotland, with funding from the Scottish government's Chief Scientist Office, the University of Aberdeen is carrying out an economic evaluation of whole genome sequencing for the diagnosis of rare conditions in Scotland. The evaluation will compare the costs of whole genome sequencing to its broader value to service users ensuring a holistic and patient centred approach is taken when measuring the benefits of whole genome sequencing. This builds on work by the Scottish Genomes Partnership – a partnership of Scottish Universities and the NHS – in collaboration with Genomics England to pilot whole genome sequencing for the diagnosis of

Scottish rare disease patients as part of the 100,000 genomes programme. The results of the health economic analysis will be used to guide the development of Scotland's future diagnostic service for rare disease patients. In addition, to support the ongoing expansion of genomics medicine in NHS Scotland, NHS Scotland National Services Division have carried out a major service review of the regional Genetic Laboratories. The review group, which consists of clinicians, services planners and representatives of other devolved administrations, have developed a number of recommendations which, when implemented will ensure NHS Scotland has the infrastructure, workforce and service capacity to react to an evolving genomics health care service

All of these are examples of healthcare systems in each UK nation beginning to transform through adoption of genomic healthcare. It is our ambition to look at current areas of difference in approach and work on how we can address these for patient benefit. For example, the Genomic Test Evaluation Working Groups established by NHS England and Improvement have been designed to bring together UK-wide expertise to collectively evaluate new genomic science and technology for genomic testing of rare and inherited diseases, cancer and for pharmacogenomics, enabling commissioning decisions to be made in our respective health services.

Pathogen genomic sequencing is another area where UK-wide collaboration is important and never more so than in our collective tackling of the COVID-19 pandemic.

At the beginning of the pandemic, in April 2020, the COG-UK (COVID-19 Genomics UK Consortium (https://www.cogconsortium.uk/)) was set up to provide a UK-wide SARS-CoV-2 genome sequencing capacity. COG-UK supported public health agencies in the analysis of SARS-CoV-2 to identify and monitor variants of concern and to track the introduction and spread of COVID-19.

Since 2021, delivery of a national SARS-Cov-2 genomics service has been led by the 4 national public health agencies working with partners including the Wellcome Sanger Institute and CLIMB-COVID. Coordination across the UK was overseen by the UK Strategic Public Health COVID-19 Genomics Advisory Board. At its final meeting the board endorsed the transition to a wider UK Pathogen Genomics Board. Work to take this forward will commence in financial year 2022 to 2023. To date, the UK has shared over 2.25 million genomes on public databases with the international community.

The UK Rare Diseases Framework, published in January 2021, is another important initiative that fosters UK-wide collaboration and outlines a national vision for how the UK will improve the lives of those living with rare diseases. As around 80% of rare diseases have an identified genetic origin, the UK's strengths in genetics and genomics have clear potential to accelerate diagnosis and improve understanding of rare conditions, thereby driving improvements in care for patients with rare disease.

Finally, while diagnosis and personalised medicine, and research form distinct pillars of Genome UK, we are clear that they cannot be implemented in isolation. Our shared principles state that health care systems and research programmes (including those funded by the medical research charities as well as industry) will work in partnership for patient benefit, and it is this interaction and partnership that leads to the many exciting and important advances in genomic research and its applications, opening up new routes for diagnosis and novel treatment opportunities. We are therefore committed that all parts of Genome UK should interact with and cross-fertilise each other to ensure high-quality research outcomes, which are already leading to improved diagnoses for UK patients, more personalised treatment and better patient outcomes.

Our shared commitments are:

- we commit to working together to build on our successful partnership on SARS-CoV-2 sequencing to develop public health pathogen genomics services across the UK. The UK Health Security Agency (UKHSA) will set up a task and finish group, with representatives from the governments and public health agencies across the UK. The group will build upon the model of the UK COVID-19 Genomics Strategic Advisory Board to develop, at pace, agreed governance to coordinate this important UK-wide activity. We will develop a core set of principles to drive the delivery of sustainable future federated pathogen genomics services, underpinned by UK-wide collaboration and co-creation
- we have committed to developing clear and tangible country-specific action plans to deliver on the collective UK Rare Diseases Framework (https://www.gov.uk/government/publications/uk-rare-diseases-framework). We will work across the UK to reduce the time in which those rare disease patients who are likely to benefit from genetic analysis receive a diagnosis, shortening what for many can be a lengthy diagnostic odyssey. While genomic analysis can be helpful, we acknowledge the need to make sure everyone in the rare disease community receives the support and care they need, including individuals who remain undiagnosed or whose rare disease does not have an identifiable genetic cause
- collectively, we will continue to review and appraise emerging genomic science (in particular the discovery of new variants and genes and their association with disease and drug response), new clinical indications and new technologies via the NHS Genomic Test Evaluation Working Groups. While the National Genomic Test Directory sets out the genomic testing strategy, which is delivered through the NHS Genomic Medicine Service in England, the Test Evaluation Working Groups are convened on a UK-wide basis to enable all 4 healthcare systems to collectively review evidence relating to different tests, allowing the devolved governments to make commissioning decisions for their own healthcare system. As part of the process to implement new genomic testing or technologies, the 4 governments will work together to understand any impact across the UK, for example where the devolved health services are currently dependent on NHS England for some testing services, in order to maintain an equitable and quality service across the UK
- over the next 3 years, the NHS Pharmacogenomics Test Evaluation Working Group, working in consultation with key stakeholders across the system including professional organisations, pharmacists and medicine optimisation infrastructure, will review and advise on how pharmacogenomics technology has the potential to reduce harmful prescribing by using genomic information to tailor prescriptions to a patient's ability to metabolise and respond to certain medications
- in cancer diagnosis and treatment, we commit to working together on the closer integration of genomic-enabled profiling of cancers with increased access to clinical trials and molecularly matched therapies, delivering on the promise of personalised medicine. For example, the Quality statement for cancer in Wales (https://gov.wales/quality-statement-cancer) requires NHS services to ensure cancer pathways use precision medicine to enable better targeting of treatments; more cases of cancer are detected at earlier stages; and realise the longer-term potential for transformational innovations, such as liquid biopsy. Personalised therapies that are already available include CAR-T (chimeric antigen receptor T cell) specialised cancer therapies, and these are available to eligible patients across the UK – for example, the NHS England Long

Term Plan (https://www.longtermplan.nhs.uk/publication/nhs-long-term-plan/) includes a commitment to offer children and young people in England access to a new generation of CAR-T. Other examples of precision cancer therapies that will benefit from this approach include epidermal growth factor receptor (EGFR) mutant non-small cell lung cancers treated with EGFR tyrosine kinase inhibitors as well as a range of other cancer mutations and their inhibitors. We will work together to explore how the offer of personalised therapies can be extended to other patient groups

- we will work together to ensure that any advanced therapy medicinal products (ATMPs) that are approved for the treatment of rare diseases are supported by the availability of appropriate clinical genomic testing across the UK. For example, as part of England's Rare Disease Action Plan, NHS England and Improvement will develop a strategic approach for gene therapies and other ATMPs by summer 2022. We will work together, including through joint horizon scanning, to share insights and inform each other's approach in this emerging field. NHS England and Improvement's initial focus will be on understanding its internal strategic approach, following which there will be engagement with key stakeholders as necessary, including with all companies developing ATMPs

Case study: collaboration on advanced therapy medicinal products

NHS Wales (via the Welsh Health Specialised Services Committee) works closely with NHS England and Improvement regarding the implementation of advanced therapy medicinal products (ATMPs) and the enabling pathways needed from the genetics service. This is achieved by NHS England and Improvement sharing their horizon scanning information with NHS Wales to inform capacity planning in Wales for ATMPs and the associated genetic services impact. NHS Wales is also well represented within the National Institute for Clinical Excellence (NICE) committee structures, including the Highly Specialised Technology Committee, which has primary responsibility for considering rare disease ATMPs, which further enhances horizon scanning and involvement in decision making. NHS Wales is also represented within the NHS England and Improvement Specialised Commissioning processes, including attendance at the Rare Diseases Advisory Group.

- we commit to sharing learning and capabilities in areas of relative strength and, where possible and appropriate, will explore cooperation in procurement to ensure maximum economy of scale. Whole genome sequencing is already being offered to specified patient groups in England and Wales and there will therefore be opportunities for collaboration between NHS England and NHS Improvement, Genomics England, NHS Wales and NHS Scotland and Health and Social Care (HSC) Northern Ireland to inform evaluation of whole genome sequencing. As part of these commitments, we will review how we work together to ensure we have the right networks in place to support quicker expansion in the use of advanced genomic tests for disease diagnosis and improve outcomes for patients across the UK

Prevention and early detection

Genome UK vision: use genomics to:

- accurately predict disease risk
- support the UK National Screening Committee to make evidence-based recommendations on relevant national screening programmes through the development of robust evidence, research programmes and stakeholder engagement

Genomics is changing the future of health and medicine, and has the potential to transform our model of healthcare from treating illness and disease to preventing illness, or detecting it at very early stages, and supporting healthy lives. Prevention and early detection are key objectives for our healthcare system – benefitting the still healthy individual through early, and often cheaper, health interventions, and also benefitting the patient at early stages of diseases offering earlier, more effective treatments. In most cases, early intervention will improve health outcomes, while also reducing treatment and care costs and helping to ensure the sustainability of the NHS into the future.

Screening is the process of identifying healthy people who may have an increased chance of developing a disease or condition, thereby allowing individuals to receive more frequent monitoring or for treatment to be initiated at an earlier stage. Genomic technologies have the potential to play an important role in screening, for example via whole genome sequencing or through the generation of polygenic risk scores, however there is work to be done to consider and address some of the ethical and privacy concerns raised by these technologies, as well as evaluating their utility in our health service. The UK National Screening Committee advises ministers and the health services across the UK about all aspects of screening and will play an essential role in appraising the viability, effectiveness and appropriateness of any new screening programmes.

We will collectively investigate the value of new genomic technologies, such as polygenic risk scores (PRS), that have the potential to identify those at highest risk of future disease and who would benefit from enhanced screening or targeted treatments and health interventions. The concept of PRS derives from genetic analyses of participants in UK Biobank, the largest and most intensively genetically and phenotypically described longitudinal cohort anywhere in the world, linking into the rich UK health record systems. PRS combines the effects of very large numbers of genetic variants to identify people who are at particularly high risk of a condition. PRS have the potential to transform public health, but many questions remain before determining whether and how they can be used routinely at scale – including the most robust disease applications for PRS and how the technology might be rolled out in the health service.

Our shared commitments are:

- given the potential for genomic technologies in the early detection of disease, we will work together to establish a clear, evidence-based position across the UK on whether and how genomic sequencing may be implemented in the newborn period, for the benefit of newborns and their parents in all parts of the UK. One opportunity for a UK-wide collaboration is the research pilot on whole genome sequencing of newborns to screen for rare genetic conditions (https://www.genomicsengland.co.uk/news/newborn-genomes-research-pilot-vision). The pilot, led by Genomics England, will also generate evidence relevant to all administrations and provide the UK National Screening Committee with high quality evidence on expanded newborn screening in the UK. The research pilot will also consider important ethical issues, such as whole-life genomic data storage, which will help to ensure that this and other genomic research is implemented in ways that maintain public trust in genomics research and its translation
- the UK National Screening Committee (UK NSC) will continue to advise ministers and health services across the UK about all aspects of population screening and support implementation of screening programmes. Following its ongoing restructure, the UK NSC will undertake work to

allow it to explore a wider range of screening programme types and it will have much closer links with research activities to support this, including greater collaboration with researchers and stakeholders

UK National Screening Committee case study for rare disease screening: from model to recommendation

The UK National Screening Committee (UK NSC) advises ministers and the NHS on screening by drawing on research and consulting stakeholders. Some rare conditions need a more detailed consultation – particularly when the science is complex and the evidence more limited. An example of this is tyrosinemia type 1 (TYR1), a very rare serious genetic condition. Newborn blood spot screening for TYR1 could potentially identify affected babies sooner, so they could be treated earlier.

The UK NSC commissioned Warwick University to build a model which compared what happens now with what would happen if screening was introduced. The model was based on an estimate of 7 babies being born each year with TYR1. Without screening, the model predicted that 4 of the 7 would be detected before symptoms develop. With screening, it modelled that all 7 would be picked up.

The UK NSC team used cohort data and information from other countries to provide evidence to support the screening pathway from the point of electronically identifying the babies to be tested, to the point of babies screening as positive and treatment outcomes. The team also worked with experts to understand how it is to live with tyrosinaemia and gain views of the benefits and harms of treatment options. These case histories were used to provide data for the model and to illustrate the consultation document.

The model concluded that screening for TYR1 would do more good than harm, but the costs per unit of additional benefit (quality-adjusted life years, QALYs) are high compared with NICE thresholds. The UK NSC is now consulting on whether it should recommend TYR1 screening given the estimated costs combined with uncertainty around aspects of the evidence. This process is one example of how the UK National Screening Committee uses modelling, expert views and consultation to provide a recommendation on whether an end-to-end screening programme, such as those based on genomics tests, is offered to patients. Delivery of screening programmes is the responsibility of the NHS in each nation.

- linked to the above commitments, we will work with national delivery partners and stakeholders to ensure that the use of genomic technologies in screening and prevention research programmes, as well as in any future clinical screening programme, are prepared for the ethical challenges posed by the use of these technologies. We will aim to facilitate this preparedness by coordinating discussions on ethics via the ‘Maintaining trust’ cross-cutting theme
- we will work together on the evaluation and review of research on genomic technologies that support early detection and prevention, to inform evidence-based and country-specific commissioning and to ensure the effective conversion of research and innovation to drive improved outcomes. Examples of this include research on single cell gene expression analysis

and the early adoption of circulating tumour DNA (CT-DNA) for early multi-cancer detection (for example, the SYMPLIFY study (https://oxfordbrc.nihr.ac.uk/new-multi-cancer-early-detection-blood-test-study-opens/)) and to inform targeted therapies earlier in the clinical pathway

- Our Future Health will be the UK's largest ever health research programme, aiming to build a cohort of 5 million volunteers from all backgrounds and from right across the UK. Our Future Health will enable pipelines to deliver integrated risk scores, that may include PRS results, to participants. UK Biobank and other studies will continue to derive new and updated PRS which can then be tested and implemented in programmes such as Our Future Health. The integrated risk scores will be piloted through the development and testing of participant-facing materials, processes and services. These will be people-centred, behaviourally-informed, and co-designed with stakeholders, including the public and participants from across the UK. Critically, the clinical, psychological, behavioural and social outcomes of providing participants with personal PRS results will also be assessed
- Our Future Health's Implementation Board will provide input into, co-ordinate and review a range of activities needed to establish and successfully deliver the programme, including the communication of integrated risk scores that include PRS results to participants. The Implementation Board will include health and care representatives on a UK-wide basis, as well as from the NHS, the National Institute for Health Research, the Office for Health Improvement and Disparities, Health Data Research UK, and professional medical societies and colleges
- NHS England and NHS Improvement will establish a UK-wide task and finish group to evaluate the use of PRS at scale in the health service. The group will jointly review the evidence on PRS across the UK and report into the existing Test Evaluation Working Groups to enable the use of evidence in individual commissioning decisions

Research

Genome UK vision: continue to lead the world in genomic research.

The UK has been at the forefront of discovery-led and translational genomics research for decades and we are home to a number of internationally leading genomic research assets. UK Biobank has sequenced the exomes and whole genomes of its 500,000 participants which represents the largest collection of genome sequences anywhere in the world, all of which are linked to participants' detailed NHS health records. Similarly, with the 100,000 Genomes Project, Genomics England holds the largest global collection of whole genome sequences from patients with cancer and rare diseases. Both UK Biobank and Genomics England are now also linking imaging data to already available clinical and genomic datasets.

The UK is also a world-leader in clinical and healthcare research – thanks to our exceptional health and care research ecosystem including the NHS, world class universities and research infrastructure (including that funded by the National Institute for Health Research in England, Health and Care Research Wales, Health and Social Care Research and Development Division in Northern Ireland and NHS Research Scotland), a strong life sciences sector, and world class medical research charities and regulators.

Our joint vision for UK clinical research delivery (https://www.gov.uk/government/publications/the-future-of-uk-clinical-research-delivery/saving-and-improving-lives-the-future-of-uk-clinical-research-delivery) highlights clinical research as the single most important way in which we can improve healthcare – by identifying the best ways to prevent, diagnose and treat conditions. The UK is already one of the top 3 destinations for delivery of commercial early phase trials and delivered 12% of all global trials for

innovative cell and gene therapies in 2020. Our combined strengths in genomics research, clinical genomics and clinical research, therefore, now offer unique opportunities to identify and approach patients who, as a result of genomic or genetic diagnosis, may be eligible for specific studies and who may in the future form part of a recallable clinical cohort for clinical trials, to discover new treatments in rare and common diseases.

The UK's investment and expertise in genomics mean that we now have an unparalleled opportunity to use genomic research assets to drive the next generation of life sciences discoveries. We will therefore work together to support post-pandemic recovery and growth in clinical research to deliver genomics-enabled clinical trials and support the growth and research and development of innovative genomics-focused companies.

In England, the Department of Health and Social Care will publish the final version of its data strategy Data Saves Lives in Spring 2022. The strategy sets out the critical role of health and research data in the transformation of the health and care sector. The current draft strategy (https://www.gov.uk/government/publications/data-saves-lives-reshaping-health-and-social-care-with-data-draft/data-saves-lives-reshaping-health-and-social-care-with-data-draft) includes commitments that will empower researchers across the UK with the data they need to develop life-saving treatments and new models of care, make progress towards bringing together genomics data assets and work with NHS England and Improvement to ensure genomic data generated through clinical care is fed back into patient records. This includes safe environments to securely analyse people's sensitive health data such as the rich genetic and genomic data hosted by Genomics England, UK Biobank and Our Future Health, alongside one of the world's most comprehensive collections of disease registries.

Another area where the UK life science sector has a unique opportunity to coordinate, collaborate and combine our expertise is functional genomics. Whole genome sequencing and other genomic tests have identified thousands of genetic variants known to be implicated in disease pathogenesis. But relatively little is known about their function and the challenge now is to understand how these variants mediate their effects, both in order to further our understanding of disease and to speed up successful drug development. Novel molecular and cell biology tools, including single cell sequencing, dynamic gene expression profiling, and systematic CRISPR, combined with insights from genomic datasets and integrated with advanced imaging and pathology, will provide opportunities for high throughput approaches to understand the role of variants and identify novel drug targets. The Medical Research Council and UK Research and Innovation partners, as major funders of discovery and translational science and research, are well placed to convene and coordinate such an initiative.

Given the rapid advances in large-scale genomic and other -omic assays, many of which utilise disruptive technologies that have been developed within the UK (such as Illumina and Oxford Nanopore), the UK is extremely well placed to take advantage of research assets that combine genomic and other -omics data at scale. With large-scale genomic and metabolomic data already available, the UK Biobank – a UK-wide and internationally renowned research asset – has the ambition to add proteomic data on 500,000 participants to characterise the molecular profile of its study participants in order to further power impactful life sciences research. In addition, its ambition to incorporate the use of long-read sequencing technologies will greatly improve the understanding of the impact of structural variation on human disease and wellness, and may additionally lead to the UK Biobank becoming the world's largest epigenetic database.

Our shared commitments are:

- through the Genome UK Implementation Coordination Group and its Research Working Group, we will identify opportunities to coordinate genomic research programmes and research infrastructure, so that synergies in activities and opportunities for collaboration, including the

better coordination and harnessing of data from clinical diagnostic testing for research, can be maximised

- working across UKRI and with stakeholders, the Medical Research Council will explore funding opportunities that could support a UK functional genomics initiative which tackles the challenges faced in the field, deepens our understanding of disease and assists the identification and validation of novel drug targets
- genomic data can be used to better identify and recruit individuals who might benefit from clinical research studies, for example via Find, Recruit and Follow up (https://www.gov.uk/government/publications/the-future-of-uk-clinical-research-delivery-2021-to-2022-implementation-plan). We will work collectively to increase access to genomics-enabled clinical trials, linking up across the UK, to fulfil the commitments in Genome UK and in the UK government's vision for clinical research (Saving and Improving Lives: The Future of UK Clinical Research Delivery) and ensure research feeds back into the clinical service to improve and transform patient care
- we will ensure action to promote diversity of genomic research data repositories, for example those established by Our Future Health, Genomics England, NIHR BioResource and the SAIL (Secure Anonymised Information Linkage) Databank to reflect the diversity of the UK's population

Ethics and maintaining trust

Genome UK vision: the UK model will be seen as a leader in strong and consistent ethical and research governance of genomic data and apply regulatory standards that support rapid healthcare innovation, and maintain public and professional trust.

Genomic data is unique to every individual. Although small genetic changes will occur in different cells and tissues in our body throughout our lives, our genome will remain our constant, unique identifier. People and patients are therefore right to demand that their genomic data is handled sensitively and securely. The possibility of creating a life-long individual genomic data resource generates distinct questions regarding who should have legitimate access to this data – and how, when and to what purpose it should be analysed, processed and communicated to the individual. Other important issues include ensuring that patients have sufficient understanding to support autonomous decision making in genomic healthcare, especially regarding what are likely to be more complex diagnosis and treatment decisions.

In implementing genomic healthcare, we want to harness the tremendous 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 regarding 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.

Working together on these ethical frameworks for genomic healthcare, we will build on our strong record of examining ethical issues in bioscience and health and in developing robust models of governance and regulation. In doing so, we can lead the world in the ethics and regulation of novel applications in genomic research and healthcare, and – most importantly – maintain the trust of patients and the public.

Our shared commitment is:

- we agree on the importance of embedding ethical considerations in both genomic healthcare policy development and programme planning and implementation, while recognising the need for more discussion on how to achieve this in a meaningful way. To begin these discussions, we will hold a series of workshops later in 2022, in collaboration with the Nuffield Council on Bioethics, involving a wide range of UK-wide stakeholders and delivery partners

Engagement, dialogue and communication with patients and the public

Genome UK vision: build and maintain trust in genomic healthcare, ensuring that patients, the public and the NHS workforce are involved and engaged in its design and implementation.

We are committed to ensuring that patients and the public are at the heart of implementing the vision in Genome UK. As we have set out in our strategy, we must empower and enable patients and the public to have confidence in the potential of genomic healthcare and help shape equitable delivery. A recent report by the Government Office for Science (https://www.gov.uk/government/publications/genomics-beyond-health) includes evidence that suggests the public can generally see the potential benefits of genomics but are also aware of its potential negative impacts on privacy. A public dialogue by Genomics England (https://www.genomicsengland.co.uk/news/public-dialogue-genomics-newborn-screening) on its whole genome sequencing research pilot came to a similar conclusion – there was broad support for such an initiative provided that the right safeguards were in place.

The DHSC Data saves lives draft strategy (https://www.gov.uk/government/publications/data-saves-lives-reshaping-health-and-social-care-with-data-draft/data-saves-lives-reshaping-health-and-social-care-with-data-draft) also recognises the need to deliver truly patient-centred care, which puts people before systems, so people will have better access to their personal health and care data and can understand exactly how it is used. People will only share their information with confidence if they feel that there are proper safeguards in place, and that those entrusted with their data will keep it safe.

An important part of empowering people is to ensure greater understanding and awareness of the benefits of genomic healthcare and allowing people to make informed choices. The COVID pandemic has raised public awareness of the power and benefits of clinical research and provided an example of the relevance and importance of concepts such as genetic variation in population health. We hope that in implementing our vision for Genome UK we can build on this awareness and interest.

Our shared commitments are:

- we commit to ensuring that the public and patients across the UK are at the heart of genomic healthcare and genomic research programmes by providing opportunities for representatives to be involved in discussions and decision-making both in healthcare genomics and genomics research. Where possible, we will involve existing patient and public representatives and groups. However, we will also explore new routes of policy co-development between experts, academics, patients and the public. Examples of successful public and patient involvement in genomic healthcare include the Genomics Partnership Wales Patient and Public Sounding Board (https://genomicspartnership.wales/sounding-board/), the Genomics England Participant Panel (https://www.genomicsengland.co.uk/patients-participants/participant-panel) and the People and Communities Forum in England. With these and other initiatives, there will likely be opportunities to explore collaboration with other genomic research participants across the UK

- Our Future Health is establishing a governance structure that has public and participant involvement and engagement embedded throughout. A Public Advisory Group has been set up comprising 14 members of the public, and this will evolve into a Participant Advisory Group in the next phase of the programme
- in addition to involving the public and participants in the governance structure, Our Future Health is also undertaking a series of other activities to ensure that the research programme is designed with the public and participants at its heart. For example, Our Future Health has commissioned its first UK national annual survey to start tracking attitudes over time, alongside convening a Citizens' Summit to explore specific issues around genomics in greater depth. Additional activities include a range of co-design workshops, interviews and focus groups
- we will also explore appropriate and specific engagement with the UK Rare Diseases Forum online platform, which hosts an active discussion space for members of the Rare Diseases community and has patient representation from across the UK around the delivery of the UK Rare Diseases Framework. Our engagement would aim to ensure that our work in genomic healthcare implementation is representative of the specific needs of the UK rare disease community while recognising that 20% of rare diseases do not have a known genetic basis
- we commit to providing appropriate and relevant information to patients and the public and work with stakeholders to ensure the impact of our respective implementation plans will be understood by those who will benefit from them

Data

Genome UK vision: deliver UK-wide, coordinated approaches to data and standardise the way in which genomic data is recorded.

Genomic data is already transforming the way in which patients are diagnosed and treated for diseases and is enabling researchers to discover the next generation of medicines and diagnostics. The UK is home to many world-leading institutions that house genomic data. However, these have tended to be developed in isolation for a specific purpose, leading to poor interoperability and difficulty in co-analysing different datasets. To implement our vision for data set out in Genome UK, we plan to link, or federate, trusted research environments (TREs), including those hosted by UK Biobank, Genomics England and Our Future Health, creating secure spaces where accredited researchers can access and securely analyse sensitive data without breaching privacy. This means that in-depth analysis can be undertaken on rich multimodal datasets, but without identifiable information ever being seen by researchers and analysts. In March 2022, initiatives to progress this work were announced in England (https://www.gov.uk/government/news/260-million-to-boost-healthcare-research-and-manufacturing).

In Genome UK we set out a number of ambitions to transform our capabilities in genomic data over a 10-year period, based on a set of agreed principles and use of shared data standards that would allow a federated approach to data sharing and improved use of AI and machine learning tools across databases.

We aim to collectively build on our successful UK-wide data collaboration during the COVID-19 pandemic, such as those pioneered by the COG-UK consortium and the UK Joint Biosecurity Centre. We also aim to work closely with units developed following the pandemic such as the UKHSA Centre for Pandemic Preparedness, leveraging connections with the WHO on their Genomics Strategy, the International Pathogen Surveillance Network (IPSN) and other bi- and multi-lateral agreements on surveillance, modelling and forecasting.

Genomic data is much more useful when combined with wider healthcare information, such as scanned pathology and radiology images, blood tests or hospital admission statistics and data initiatives such as the SAIL (Secure Anonymised Information Linkage) database in Wales are already aiming to achieve this. When combined, this data allows researchers to make more informed links between genetic changes and disease development, improving the accuracy of diagnosing genetic conditions and providing a platform to launch drug discovery programmes. This kind of analysis is accelerated by the latest developments in artificial intelligence and machine learning.

Case study: SARS-CoV-2 data sharing and exploitation as part of the pandemic response

Across the UK, multiple pathogen genomics services already exist, delivered at varying levels across England and the devolved administrations. Many of these services have ISO 15189:2012 medical laboratory accreditation, and all are dependent upon digital infrastructure to enable the generation of actionable information from sequence data. These services often exist in silos, creating barriers to sharing approaches and data across the UK, creating inequalities of service across the UK because of the current digital systems that exist to provide current services.

In contrast to existing pathogen genomics services, the sequencing response to the COVID-19 pandemic has seen unprecedented co-creation across the UK, and across healthcare/public health, academia and government. In response to an urgent need, a complete analysis platform for UK SARS-CoV-2 sequence data, CLIMB COVID, was built in less than 3 days in March 2020. This platform was put in place to provide a single data sharing and analysis platform which would bring together all UK SARS-CoV-2 genomes, wherever they were generated, and enable their analysis in real time. Providing real-time analysis on a single, combined UK dataset has enabled the generation of actionable intelligence at multiple scales – ranging from outbreak analyses for infection prevention and control in hospitals, up to information on the shape and progression of the pandemic across the UK, to inform government policy. Collectively CLIMB COVID currently stores and analyses over 2 million SARS-CoV-2 genomes from across the UK, with the data and analysis outputs being actively used by the 4 UK public health agencies as well as numerous NHS trusts.

The exploitation of pathogen genomics data as part of the COVID-19 pandemic paints a picture of what is possible in a future where pathogen genomics data is rapidly shared across the UK as required. With shared data and common analysis approaches, expertise can also be more effectively pooled and analyses can be undertaken more collaboratively across the UK public health agencies for the benefit of patients and the public. The federated model also means that each nation is able to use the data to meet their local needs.

The success of the SARS-CoV-2 genomics efforts in the UK has been a federated approach to sequencing and analysis underpinned by a multi-node data processing infrastructure which works to standardise analysis and enables work to be undertaken on a UK-wide basis. This infrastructure, underpinned by data sharing agreements that span the UK public health agencies, provides a validation of a future federated approach, and demonstrates the enormous potential that exists through working collectively across the UK to improve how we link, combine and use our genomic data.

Our shared commitments are:

- over the next 3 years, we will support research initiatives to test the challenges and develop solutions to federated approaches to data access. Led by the Genome UK Implementation Coordination Group Data Working Group, this work would look to link datasets from across the

UK, and federate where appropriate, with potential examples including the Trusted Research Environments hosted by Genomics England, UK Biobank and Our Future Health as well as secure data environments hosted by SAIL (Secure Anonymised Information Linkage), the All Wales Medical Genomics Service (AWMGS), the Congenital Condition and Rare Disease Registration Information Service for Scotland (CARDRISS), the Northern Ireland Genomic Medicine Service, and The National Congenital Anomaly and Rare Disease Registration Service (NCARDRS). Some of this will also build on work under the UK Rare Diseases Framework to align and standardise these national rare disease registries

- we will collaborate in our implementation of our wider data strategies over the next 3 years – for example, in England the draft data strategy Data saves lives (https://www.gov.uk/government/publications/data-saves-lives-reshaping-health-and-social-care-with-data-draft/data-saves-lives-reshaping-health-and-social-care-with-data-draft) is already available and the final version will be published in Spring 2022. In Northern Ireland a data strategy is in development. Closer collaboration will ensure that the sharing of best practice in how our data is stored and accessed extends beyond genomic data, allowing a deeper potential for federating our datasets and a greater shared opportunity to deploy AI and machine learning tools across organisational boundaries
- it is crucial that we have continued collaboration across the UK to implement our ambitions for genomic data in a coordinated and detailed manner. As such, the NHS Transformation Directorate (formerly NHSX) will convene a UK-wide genomic data working group with representation from experts across the UK's genomic healthcare landscape to provide a forum for coordination and collaboration and to ensure that our processes maintain public trust in the safe, appropriate, and responsible use of personal data for both clinical care and research
- one clear action for this UK-wide group is to build a collective understanding of the barriers for sharing genomic healthcare data across UK borders and health agencies, as well as agreeing the principles and interventions to overcome these challenges. These interventions could range from increasing trust and transparency on a UK-wide basis via collaboration and relevant policy levers, to necessary changes through legislation to remove unnecessary data sharing barriers
- we will utilise UK-wide fora, such as the National Genomics Board, the Genome UK Implementation Coordination Group Ethics workstream and others, to discuss the wider impacts and uses of genomic data beyond our health programmes, such as the ethical questions around using genomic data over a lifetime. Careful consideration needs to be given at a UK level to appropriately plan for the future via guidance and legislation, and the National Genomics Board will work to ensure these issues are appropriately considered, alongside work led by GO-Science and wider government

Workforce development

Genome UK vision: support and enable healthcare staff to deliver the benefits of genomics by training and supporting them to acquire the relevant knowledge and skills, and developing clinical pathways and standards of care.

The ambition of the Genome UK vision cannot be achieved without ensuring that the workforce have the necessary skills and knowledge to deliver genomic healthcare. Staff need to understand how genomic tests fit into clinical pathways, identify which patients require which type of test, and be able

to interpret and communicate the results of these. For UK patients to receive the benefits of the latest advances in genomic technology and infrastructure, we need a workforce that develops in parallel, so that we empower healthcare professionals with the confidence and up to date knowledge needed to deliver these innovations. This will involve embedding formal education in genomics into speciality training programmes, as well as providing clinical staff with the resources to stay up to date with the latest advances in the area and utilise the available clinical pathways. By creating a National Framework for genomics education, we can ensure consistency of capability across the UK.

Key delivery partners in this work are Health Education England (HEE) and its Genomic Education Programme, which supports the NHS Genomic Medicines Service and ensures that the 1.2 million-strong NHS workforce has the knowledge, skills and experience to keep the UK at the heart of the genomics revolution in healthcare, and the Academy of Medical Royal Colleges, which sets the standards for how doctors are educated, trained and monitored through their careers.

Our shared commitments are:

- we will work together to establish a joint workforce group across the UK to share best practice and strategic priorities
- to keep pace with rapid advances, we are committed to supporting clinicians across the UK in understanding and adopting genomic medicine. This includes training for healthcare professionals and providing them with the resources they need. Health Education England will develop and launch the testing phase of their educational resource ‘GeNotes’ which will include a Knowledge Hub that is accessible to clinicians across the UK
- GeNotes facilitates the use of the test directory and integration of genomic medicine across the healthcare specialities and professions. The Health Education England Genomics Education Programme (GEP) will establish a UK-wide working group with representatives from each devolved government to discuss how GeNotes and other GEP resources can be made accessible and applicable to healthcare settings across the UK
- the Academy of Medical Royal Colleges, as the coordinating body for the UK and Ireland’s 24 medical royal colleges and faculties, will continue to engage its members and others on UK-wide coordination of genomics education, implementation and workforce issues
- in addition, the Academy of Medical Royal Colleges, in partnership with HEE and NHS England and Improvement, will continue work on developing and implementing a national framework for genomics education, which will involve:
- leading implementation of a general genomics curriculum for post-graduate medical education that would be suitable for tailoring to other professionals
- leading implementation of a framework for clinical pathways involving genomics with educational resources needed along those pathways
- promoting a network of national, regional and local genomics advisors with an accompanying framework of competencies
- providing overview and approval of documents and policies to ensure genomics is optimally implemented across the healthcare landscape
- providing a mechanism for communication between central bodies and stakeholder organisations across the UK

Industry

Genome UK vision: make the UK the best location globally to start and scale new genomics healthcare companies and innovations, attracting direct investment in genomics by the global life sciences industry and increasing our share of clinical trials in the UK.

In July 2021, the government and the life sciences sector published its Life sciences vision (https://www.gov.uk/government/publications/life-sciences-vision). The vison sets out the government's and the sector's collective ambition for the UK to build on the scientific successes and ways of working, from COVID-19 to tackling future disease challenges (including cancer, obesity and dementia), ageing, secure jobs and investment and become the leading global hub for life sciences.

The vision recognises that to remain competitive in the life sciences and deliver on its ambition, the UK will need to focus relentlessly on areas in which it already has, or can gain, a competitive advantage – such as genomics and health data. It also recognises that, for genomics, our ambition needs to be to create scale. We already have fantastic expertise, tools and world-leading initiatives in UK genomics – our challenge is how to bring these together in a way that is transformative and places the UK firmly ahead of its competition, while making it a valued partner for international collaboration and an attractive location for investment.

This can be achieved through working closely with the sector on the existing and planned initiatives included in these shared commitments, such as enhancing our genomic UK-wide research infrastructure, large pilot studies to evaluate variants and their role in predicting disease risk, evaluation of new genomic tools for early disease detection, new tools for improved cancer diagnosis and delivering a world class offer to support functional genomics studies.

The UK has a diverse industrial life sciences sector, comprised of large multinationals, SMEs and spinouts – all of which bring unique value and expertise to the UK's genomics ecosystem. The innovations coming from the commercial UK genomics sector and their international partners and collaborators will underpin developments in research and healthcare for years to come. It is vital that we capitalise on the UK's existing strength by continuing to foster an environment that allows companies to develop new treatments, deliver effective innovations to patients and grow at scale.

Our shared commitments are:

- we will work closely with industry as we deliver these UK-wide shared commitments. Our first action will be to convene a joint workshop in partnership with trade associations, including the Association of the British Pharmaceutical Industry (ABPI) and Bioindustry Association (BIA), in spring 2022 to discuss industry priorities and how these should be reflected in Genome UK implementation
- we will encourage innovative and cutting-edge industry partnerships in research and development across the UK to support implementation of Genome UK
- we will foster a supportive and attractive environment for genomics SMEs, by encouraging access to data assets, biosampling capabilities, and collaborative academic and clinical expertise across the UK
- we will continue to work with industry and charity partners on initiatives such as Our Future Health and UK Biobank

Conclusion

In our Genome UK strategy, we set out, for the first time, a comprehensive and ambitious vision for the future of genomic healthcare: a future where genome sequencing, genomic tests and integrated genomic and other health data will help to detect the risk and very early stages of disease to support

early intervention, and where genomic and other -omic technologies can speed up diagnoses and support the development of better, more precise treatments for many diseases, including cancer.

Here we commit to following through on this vision – by working together we will achieve better coordination and collaboration on genomic healthcare for the benefit of patients across the UK, while recognising the differences in our respective healthcare systems and structures. Our commitments will strengthen our ability to share expertise and establish new collaborations and partnerships to progress genomic healthcare not only across the UK but worldwide, securing our global leadership in genomics and the wider life sciences.