Partneriaeth Genomeg Cymru
Genomics Partnership Wales

Genomics Delivery Plan for Wales (2026-2029) – Draft Actions

Contents

Introduction

This draft document contains the **planned actions** for genomics for the next three years, for consideration and feedback from a wide range of audiences.

The final Delivery Plan will also include the following additional information:

- Forewords from Welsh Government representative, Senior Responsible Officer for Genomics Partnership Wales and Patient and Public Sounding Board
- Evidence of the impact of involving our patients in genomics work
- The story so far – Genomics Partnership Wales's remit and purpose, achievements and challenges to date
- Strategic landscape in Wales and the UK
- Genomics ecosystem, key drivers and delivery themes for strategic approach
- An appendix table detailing the metric, impact/benefit and financial arrangements for each action

Planned delivery activity for the Genomics Partnership Wales programme is indicated by specific actions in this Plan or via other genomics plans (links are provided in appropriate sections).

Action owners are shown *(in italics)* after each action – please note that other stakeholders may be involved in the activity, but the owning organisation will lead the work. Acronyms as follows:

GPW – Genomics Partnership Wales

WGP – Wales Gene Park

AWMGS – All Wales Medical Genomics Service

PHGP – Public Health Genomics Programme

DHCW – Digital Health and Care Wales

LSHW – Life Sciences Hub Wales

WG SRE – Welsh Government, Science, Research and Evidence Division

Delivery Theme 1: A Focus on People

Workforce and Training

To fully realise the benefits of genomics within healthcare, it is essential that that the workforce across many different clinical and public health services are equipped to use genomics in their practice. Genomics Partnership Wales and Health Education and Improvement Wales have developed a comprehensive, strategic plan that provides a roadmap of activity from 2025 until 2028. This workforce plan consists of actions that will drive genomic literacy skills across all aspects of healthcare and clinical practice. It also addresses current challenges and future requirements of the specialist genomics workforce to ensure the skills and scale are in place. The overall aim is to ensure that the opportunities presented by genomics are realised for the benefit of our patients and the Welsh population.

Genomics Strategic Workforce Plan

Patient Involvement

Genomics Partnership Wales will build on the established involvement framework of the Patient and Public Sounding Board to strengthen and broaden the utility and impact of working with the public to deliver genomics activities.

ACTION 1.1: Achieve equitable representation of the population on the Sounding Board through broader engagement with under-served community groups *(GPW)*

ACTION 1.2: Ensure patient and public involvement in the delivery of all strategic genomics activities outlined through quarterly consultation sessions, representation on specific groups as appropriate and other specific forums as appropriate *(GPW)*

ACTION 1.3: Develop an evaluation strategy for the genomics programme that is co-produced and monitored by the Patient and Public Sounding Board to demonstrate the impact and benefit of Genomics Partnership Wales activities *(GPW)*

Engagement and Communications

With the recognised importance of genomics in medicine, there is an increasing need to:

- Educate health professionals to integrate genomics into clinical care
- Engage the public and patients in understanding the implications of genomics in health
- Ensure patients and families affected by rare and genetic conditions are involved in policy development and research
- Enthuse and encourage the next generation to explore careers in genomics

Wales Gene Park plays an essential role in addressing all the above through their genomics education, engagement and patient involvement activities across Wales. By aligning its initiatives with national policy and leveraging partnerships, its model for effective genomic engagement is one that is responsive to the needs of all stakeholder groups. With recognised expertise and a long, successful track record, Wales Gene Park's Education and Engagement team will continue to underpin the ambition of the Partnership, delivering an inclusive, collaborative and equitable multi-faceted work programme tailored to different target audiences; it will:

- **Improve** professional knowledge and understanding of genomics and **increase** interest in related careers, in alignment with the *Genomics Strategic Workforce Plan*. Additionally, **integrate** effective patient and public involvement and engagement in research through continued support and facilitation of opportunities within the genomics research community in Wales (*Strategic Research Plan for Genomics in Wales*)
- **Inspire** young people to learn about genomics and be attracted to pursuing education and careers in the field, as defined in the *Genomics Strategic Workforce Plan*

ACTION 1.4: Create and implement annual plans for Wales-wide events that bring together health professionals, researchers, patients and the public, to showcase genomics and highlight advances in Wales and beyond *(WGP)*

- **Interest** and **inform** the Welsh public about genomics to continue to raise awareness and help cultivate a genomically-informed society

ACTION 1.5: Create and implement a broad programme of established and new engagement activities with the public, integrating new technologies to support delivery as appropriate *(WGP)*

- **Involve** those with lived experiences of rare, genetic and undiagnosed conditions, and **include** the voices of underrepresented communities, to effectively **influence** and **inform** genomics policy, research and service delivery in Wales.

ACTION 1.6: Provide key focuses (screening, rare disease research opportunities, inequities in rare disease care and treatment, health professional awareness, care coordination) into UK Rare Disease framework priorities following its conclusion in January 2026 *(WGP)*

ACTION 1.7: Collaborate with key partners in the Senedd to highlight genomics through Cross-Party Groups, drop-in session participation and marketplace exhibitions *(WGP)*

ACTION 1.8: Support innovation within healthcare through initiatives such as the Welsh Rare Care Centre *(WGP)*

Genomics Ambassadors are individuals who have previously been members of the Patient and Public Sounding Board; they want to remain involved in the programme and can use their Sounding Board and personal experience to promote genomics within communities in Wales, to strengthen the engagement needs to support the Genomics Partnership Wales programme.

ACTION 1.9: Develop a comprehensive suite of materials and framework to support and fully utilise the Genomics Ambassadors to increase public awareness and interest in genomics, ensuring that engagement plans complement and strengthen the activities led by Wales Gene Park *(GPW)*

Communication activity is a critical cross-cutting theme within our genomics ecosystem and underpins every aspect of delivery. We will build on our strong communication activities to date, take an all-Wales approach collaborating with all areas of the Partnership and engage with audiences using the most appropriate channels to amplify our key messages.

ACTION 1.10: Deliver a robust communications approach that is evidence-based and appropriately tailored to different audiences, strengthening partnership communications in the following ways:

a) Improve communication with non-traditionally engaged groups to ensure equitable representation of patient voice, and access to health care information and resources

b) Engage the wider public and health professionals on genomics with use of appropriate language, myth-busting information and accessible materials that show the benefits and relevance of genomics to them

c) Emphasise the importance and impact of genomics through case studies and lived experience by promoting the contribution of the Genomics Partnership Wales Sounding Board and Genomics Ambassadors, and showcasing our patients' stories

d) Ensure alignment across the broad genomics landscape in Wales to identify opportunities for increasing the visibility of the work nationally and internationally *(GPW)*

ACTION 1.11: Review and develop the Genomics Partnership Wales website to ensure appropriate, current information and resources to cater for several different audiences *(GPW)*

Delivery Theme 2: Genomic Service Delivery

The number and range of genomic services delivered in Wales to individual patients and at a population level has grown exponentially in recent years. Genomics offers significant opportunities to improve healthcare in Wales through the provision of new capabilities built on a foundation of cutting-edge science, increased understanding and the co-ordinated approach that has been established through Genomics Partnership Wales. This rapid growth presents us with a challenge; we must be forward looking, whilst focusing resources on efficient and timely service delivery, ensuring our operational performance meets our patients' needs.

Our vision for genomics services over the next three years will see a continuation of implementation of new services, alongside the consolidation and improvement of our existing services – all done to ensure equity of access to the benefits that genomics brings across Wales.

Genomic testing services in Wales and alignment to NHS England

Genomic testing enables, informs and underpins a range of health services including earlier and more accurate diagnosis, personalised treatment, prevention, reproductive decision support, infectious disease management and health protection. It is imperative that genomic test results are integrated with wider health data to enrich patient and population information, and to form the basis for evidence generation, service development and policy action.

Unlike the English system, in Wales key delivery functions with respect to genomics for clinical testing, population and public health are provided by two NHS organisations.

To ensure equity of access for Welsh patients, the suite of human genomic tests available in Wales are provided by All Wales Medical Genomics Service and align to the commissioned NHS England Cancer and Rare Disease Test Directories. The testing in England is provided by NHS Genomic Medicine Service on behalf of NHS England Specialised Commissioning and concentrates on the provision of rare disease, cancer and population genomics (including pharmacogenomics) testing, with emphasis on technology and innovation. The value-based approach in Wales centres around clinical utility and information necessary to support patient care, in line with the principles of Prudent Healthcare. In this context, the exact delivery of the testing performed may vary from NHS England; for example, there may be a difference in the testing panel or methodology used.

Across the UK there is no pathogen genomic test directory. The suite of pathogen genomic tests available in Wales are provided by the Public Health Wales Pathogen Genomics Unit, and in Wales the implementation of tests is driven by clinical requirements established and managed through the Public Health Genomics Programme, which integrates the needs of infectious disease and microbiology clinicians, and the needs of the Health Protection teams.

Pillar 1: Cancer Genomics Services

Cancer care and improving outcomes for patients remains a priority in Wales; the use of innovative genomic technologies is key to providing faster and more accurate diagnosis, enabling rapid access to precision treatments for cancer patients. Supporting further use of routine cancer genomic data to enhance recruitment to clinical trials is a priority. The introduction of standardised and timely (including urgent) testing ensures equity of access to diagnostic testing irrespective of postcode across Wales. All Wales Genomics Oncology Group will support cancer genomics by further

developing and strengthening established links between All Wales Medical Genomics Service clinical and laboratory services, oncologists, pathologists and haematologists in Wales. Bringing all partners together by recognising each other's strengths and weaknesses, is key to delivery of these new technologies and services across Wales.

ACTION 2.1: Deliver equitable cancer testing aligned to NHS England Cancer Test Directory, including the phased implementation of clinical cancer trial targets; deliver value for money, develop greater expertise and expand current innovative applications to include methylation arrays, signature profiling (e.g. Homologous Recombination Deficiency), long read sequencing and liquid biopsy *(AWMGS)*

ACTION 2.2: Provide a coherent approach to improve turnaround times for blood and solid cancer testing aligned to professional and published guidance. NHS Wales will be required to meet the recent NHS England's National Cancer Board (2023) approved 'new' end-to-end turnaround times for cancer patients where the combined histopathology and genomic results are required by day 7 (urgent), day 14 (routine) or day 28 (for future clinical decision-making), from the time of sample acquisition. It is recognised that joint working is required across national pathology, genomics and cancer programmes to achieve these targets *(AWMGS)*

ACTION 2.3: Develop a plan to ensure that every cancer patient as appropriate receives comprehensive genomic analysis and molecular profiling at diagnosis (tissue or liquid biopsy) to enable relevant clinical care and management. The list of genes will be identified from standard of care DNA and/or RNA panels from large gene panel, whole exome sequencing or whole genome sequencing testing *(AWMGS)*

ACTION 2.4: Deliver a somatic Whole Genome Sequencing service for a sub-set of defined haematological (blood) and solid cancers, including a strategy for the implementation of a fresh frozen tissue pathway. Most cancer tests delivered are next generation sequencing panels and other targeted technologies, and these tests obviate the requirement for whole genome sequencing for all cancer patients (*AWMGS*)

ACTION 2.5: Establish an NHS Wales Genomic Tumour Advisory Board to support clinical teams across Wales with the interpretation of genomic test results and provision of information about possible appropriate clinical trials or treatments *(AWMGS)*

ACTION 2.6: Generate further evidence to adopt liquid biopsy testing for cancer into commissioning through nationally agreed research projects to support earlier access to personalised treatments and clinical trials, building on the strong foundations established by the QuicDNA project since 2020. Extend the implementation of liquid biopsy for other cancer indications (for example, Cancer of Unknown Primary) *(AWMGS)*

Pillar 2: Inherited Genomics Services

Inherited genomics services cover all rare, inherited diseases, including those predisposing to intellectual disability, congenital anomalies, neurological disorders, congenital heart disease as well as inherited cancer predisposition, and pharmacogenomics. All Wales Medical Genomics Service aims to provide diagnoses or confirmation of suspected genomic conditions, prediction of risk for family members, recommendations regarding clinical management, surveillance, treatment and prognosis. The All Wales Medical Genomics Service clinical team provides specialist advice to other clinical areas through attendance at various multi-disciplinary team meetings and plays an important

role in the education of the non-specialist workforce about genomics; it also takes the lead in management of very complex, rare diseases with multisystem involvement that do not naturally align with any other medical specialty. This wide range of services are for patients of all ages from prenatally through childhood, adolescence, adulthood to end of life

Alignment to NHS England genomic testing services

The NHS England Rare Disease Test Directory includes testing for rare and inherited disease, including inherited cancer, and pharmacogenomics; these areas are included in this **Inherited Genomics Services** section

Pharmacogenomic testing has the potential to revolutionise healthcare by tailoring treatments to individual genetic profiles, leading to more effective and safer use of medicines.

The *Pharmacogenomics Delivery Plan for Wales* will ensure that a strategic approach is taken to the development of pharmacogenomic services across community, primary and secondary care in line with the publication of services on the NHS England Test Directories and/or strategically funded programmes.

ACTION 2.7: Work with the *Rare Disease Implementation Network* to align to the aims and actions in the new *Rare Disease Action Plan* in both Wales and UK. This includes a critical requirement to review the Wales Community Child Health Network guideline genomic testing strategy to deliver an equitable, value for money rare disease diagnostic pathway that is also aligned to the NHS England Rare Disease Test Directory *(AWMGS)*

ACTION 2.8: Continue to be the single whole genome sequencing provider in NHS Wales that delivers equitable rare disease testing aligned to NHS England Rare Disease Test Directory, delivering efficient diagnostic pathways while developing greater expertise and expanding current testing applications. This includes evaluating the application of whole genome sequencing within diagnostic pathways *(AWMGS)*

ACTION 2.9: Introduce new technologies and bioinformatic analysis tools (including artificial intelligence) that have been evaluated to improve clinical outcomes *(AWMGS)*

ACTION 2.10: Review the NHS England Foetal Anomaly Gene Panel service and understand the utility to urgently inform the clinical management of pregnancy alongside other prenatal testing *(AWMGS)*

ACTION 2.11: Continue to repatriate specialist services from external providers to provide a value for money streamlined service with improved turnaround times with increased internal specialist clinical expertise *(AWMGS)*

ACTION 2.12: Support evidence generation and operational requirements for the reporting of increased pharmacogenomic targets from reactive to pre-emptive testing generated through rare disease and other testing. Work towards automated, high throughput pharmacogenomic testing, requiring minimum scientific input, including exploring opportunities with other UK nations to deliver at scale and pace *(AWMGS)*

ACTION 2.13: Identify individuals at high risk of adverse drug reactions. Implement integration of pharmacogenomic profiles into integrated NHS healthcare pathways linked to NHS England's Test Directory *(AWMGS)*

ACTION 2.14: Establish an Inherited Cancer Predisposition Register for patients in Wales with a proven genetic predisposition to cancer, which can link with screening services to ensure these cohorts of patients are offered appropriate, timely surveillance, leading to earlier detection and improved outcomes for those who are diagnosed with cancer (*AWMGS*)

ACTION 2.15: Establish a formal Inherited Cancer Predisposition Management service to support patients following a diagnosis of a genetic predisposition. This will include referral for surveillance in accordance with national guidance, advice about risk management options including risk-reducing surgery where appropriate, support with reproductive decision making, psychological support and advice about targeted treatment options where these are available (*AWMGS*)

ACTION 2.16: Develop an Inherited Cardiovascular Conditions service to provide equitable care and appropriate regular surveillance for patients, both children and adults, at risk of cardiovascular diseases across Wales (*AWMGS*)

Neuropsychiatric and dementia services

All Wales Psychiatric Genomics Service was established in 2022 and is the first multidisciplinary clinical service of this kind in the UK. The service initially served adults with mental health conditions, particularly psychosis, or people with copy number variants associated with mental health conditions. The service has developed to include experts from child and adolescent psychiatry as well as psychiatric nursing, broadening the range of patients that can be seen to include children and adolescents, improving genomics literacy amongst NHS Wales mental health staff and developing new strategic direction in consultation with patients and their families.

ACTION 2.17: Strengthen genomic referral pathways for patients from mental health services, and other services, including paediatrics and intellectual disability psychiatry *(AWMGS)*

ACTION 2.18: Working with the NHS, patients, families and third sector organisations to develop referral and health monitoring pathways for those at high genetic risk of severe mental illness, to move towards a paradigm of preventative care *(AWMGS)*

ACTION 2.19: Develop resources, co-produced with individuals with lived experience of mental health conditions, and third sector organisations, to educate and support patients and their families and carers *(AWMGS)*

ACTION 2.20: Develop clinical guidelines for investigating, monitoring and treating mental health and physical health outcomes of people with identified genetic risk *(AWMGS)*

ACTION 2.21: Lead and participate in knowledge exchange activities and sharing of best practice with other clinical services, to develop the service as a leading centre of expertise in clinical psychiatric genomics in the UK and internationally *(AWMGS)*

ACTION 2.22: Support patients to participate in research to deepen overall understanding of genomic contribution to mental health problems, and create frameworks to use findings directly to improve clinical care, including treatment decisions and monitoring (the service will be used as a testbed for developing the use of routine clinical data and samples for clinical research in partnership with patients, the NHS and academics aligned to the development of the All Wales Medical Genomics Service Research and Development Strategy and in response to the *Strategic Research Plan for Genomics in Wales* *(AWMGS)*

ACTION 2.23: Support the education of NHS staff in psychiatric pharmacogenomics and the integration of evidence-based pharmacogenomic testing in mental health services *(AWMGS)*

ACTION 2.24: Genomics specialists will strengthen networks with teams caring for patients with dementia through education and collaborative working to improve access to genomic testing *(AWMGS)*

Pillar 3: Population Health Genomics Services

The development of population health genomics services will improve our ability to prevent and detect disease. Genomics is the essential tool that will enable Wales to address many of its long-term health challenges. Genomic data offers the ability to work from patient to population with transformational possibilities across the health and social care system. NHS services have a key role in generating evidence, advice and guidance to inform policy and practice, and develop and deploy clinical services that empower individuals to improve their own health while enabling earlier detection of disease.

Activity varies across the UK, and much of the key work in developing evidence in this area will be achieved through UK-wide collaboration. With the completion of large-scale UK-wide studies, the next three years will see significant work to conceive, plan and implement services based on advances from research.

A Healthier Wales highlights the need to move towards greater prevention and early intervention, to improve people's health and wellbeing, and support the sustainability of our health care system. Recognising that genomics could be a cornerstone technology in realising a more sustainable system, our plans for genomics population health revolve around building and expanding on existing clinical and public health services, while also planning for the increased use of genomic data for population health over the next decade.

On a population level, Public Health Wales has responsibility for prevention programmes, disease registries, and screening programmes. Public Health Wales works closely with All Wales Medical Genomics Service who provide the genomic testing for the screening programmes.

Population health plans are under development in England; the challenge for Wales will be to match this activity to ensure equity of access for patients, while also building on areas of Welsh strength to lead where appropriate. With an increased emphasis on population health programmes, we will build on existing partnership working to ensure that Wales sees the full benefit of new genomics services.

Public Health Genomics

Public Health is often defined as the science and art of preventing disease, prolonging life and promoting, protecting and improving health through the organized efforts of society. In public health genomics, we seek to effectively and responsibly use genomic data to deliver and inform public health services. Genomics enables the delivery of precision public health, to provide the right intervention to the right population at the right time. The potential value of genomic data is immense; it can be used in many ways to address the health challenges in Wales and benefit our population.

To deliver precision public health, we must improve our ability to understand and anticipate the health needs of our population (requiring an improvement in how we deliver public health surveillance), while also utilising tools such as genomics to improve population-wide services. We must develop evidence to enable the implementation of genomics into routine public health services, such as screening and prevention programmes. This also includes the integration of pharmacogenomics into public health surveillance systems to enhance population health outcomes, in alignment with the *Pharmacogenomics Delivery Plan*.

What is Public Health Genomic Surveillance?

The amount of genomics data generated routinely in the NHS in Wales is significantly increasing. This provides an opportunity to further embed genomics into public health surveillance. The provision of timely, accurate, actionable data relating to the health of the population is a cornerstone of realising precision public health.

The term 'surveillance' can have sinister connotations; surveillance in public health is generally designed to avoid the identification of individuals, and the purpose of the activity is to look at our population. By using aggregated information, we can derive insights to enable the delivery and improvement of population-wide healthcare services.

The World Health Organisation defines public health surveillance as 'the continuous, systematic collection, analysis and interpretation of health-related data'. In Wales, Public Health Genomic Surveillance already forms an essential part of health protection efforts to fight infectious disease and is emerging as a key tool in prevention and in the fight against chronic and environmental disease. It is used for both infectious and non-infectious disease, providing a single framework for understanding and improving the health of the population.

Public Health Genomics Surveillance will enhance our public health capabilities by:

- Detecting signals across the population that in turn enable proactive, preventative action to safeguard the health of the people of Wales
- Underpinning health protection efforts to fight infectious disease, through the detection and characterisation of outbreaks. This capability also provides early warning of impending outbreaks that could become public health emergencies
- Providing the basis for monitoring and evaluating the impact of interventions, to demonstrate their effectiveness
- Providing a better understanding of our population, enabling and informing changes in policy and guidance across all areas of public health activity. Using genomics we can improve priority-setting, planning and evaluation of public health policy and strategies
- Supporting, improving and better targeting prevention initiatives, enabling an expansion of precision public health in Wales

Public Health Genomics Surveillance potentially unlocks a host of new services, and may provide significant information to refocus, refine or revolutionise existing activities. By embedding genomics across the spectrum of public health activity, we will be better able to address the many challenges the healthcare system in Wales faces over the next decade.

Only by covering both surveillance and service development, that encompasses both screening and prevention, can we meet the ambitions in *A Healthier Wales*.

What is Prevention?

Prevention in public health involves actions to prevent a disease ever manifesting. Within Wales, health challenges such as obesity have enormous implications for our healthcare system. To ensure that the NHS is affordable in the future, it is vital that we are able to move from a system that is focused on providing acute care, to one that prevents disease from occurring, and in doing so enables our population to live longer, healthier and happier lives.

Genomics will have a significant role to play in prevention in the future; many prevention programmes are targeted at known risk groups, but with genomics there is the potential to refine these programmes, provide better stratification of the population and better target the people who need them most.

There are several successful programmes in Wales (such as All Wales Diabetes Prevention Programme, which has reduced the risk of developing Type 2 Diabetes in patients by 23%), but genomics offers the potential for further improvement, in addition to the opportunity to implement new programmes. Despite efforts around prevention, there are instances where individuals will still develop a disease. Wales currently operates a suite of screening programmes, to enable the earliest intervention possible. Genomics potentially offers new capabilities that could enhance existing screening provision (e.g. through testing for additional conditions) or could form the basis of new screening programmes.

ACTION 2.25: Evaluate the potential uses and the need for guidance in relation to population-wide genomics and insights from large-scale studies such as Our Future Health. From this, as required, support Welsh Government in enacting changes to legislation and policy that enable the widespread and routine use of tools such as polygenic risk scores within the healthcare system in Wales *(PHGP)*

ACTION 2.26: Develop a plan to extend the utility of Public Health Wales-hosted registries to make better use of the genomic results they hold, integrating this planning into new areas, including the development of a formalised Adult Rare Disease Register *(PHGP)*

ACTION 2.27: As part of the Public Health Wales Digital and Data Strategy, implement a framework that enables the automated use of genomics results as part of routine and statutory statistics and monitoring *(PHGP)*

ACTION 2.28: Utilise the Public Health Wales behavioural science programme to evaluate the communication of individual and population-based risk information based on genomic data (including polygenic risk scores), and how this could be employed in prevention and screening services *(PHGP)*

ACTION 2.29: Evaluate and plan for the integration of genomics in public health prevention initiatives, priorities and policies, with an aim to mainstream the use of genomics as a core part of public health practice in Wales. This work will provide the basis for the future use of genomic data in the delivery of population-wide programmes focused on prevention and will include using both pathogen and human genomic results generated by Welsh laboratories to identify and implement new services, and/or guidance/policy. Initial areas of focus include:

a) Integrating genomic results and data into activities focused on influencing the wider determinants of health and supporting healthy behaviours

b) Using genomic insights and results to enhance and improve the Cardiovascular Disease Prevention Delivery Plan and to support the prevention of cardiovascular, stroke and atrial fibrillation risk

c) Integrating genomics into planning as part of the Prevention-based Health and Care Action Plan and the Public Health Approach to Primary and Community Care by 2035 in Wales, which emphasises data-driven decision making

d) Integrating genomics into the activities of All Wales Diabetes Prevention Programme, using population-level data, and planning for future cases where genomic testing outputs (such as polygenic risk scores) could be used to inform diabetes prevention activities *(PHGP)*

ACTION 2.30: In collaboration with Welsh Government and NHS Performance and Improvement, support the development of systems to improve the routine postmortem use of genomics for sudden infant death syndrome and sudden unexpected death in childhood to provide comfort and certainty to grieving families, and support efforts to prevent potential sudden infant or childhood fatalities in siblings *(PHGP)*

ACTION 2.31: Develop a plan to evaluate the use of epigenetics and genetic markers of environmental hazard exposure as part of all-hazards surveillance activity within Wales *(PHGP)*

ACTION 2.32: Support and engage with the UK National Screening Committee and large studies (such as the Generation Study) to identify potential improvements to existing or future population-wide genomic services, to inform and advise Welsh Government, and to ensure that All Wales Medical Genomics Service and Public Health Wales can plan and prepare for implementation and adoption when required *(PHGP)*

ACTION 2.33: Undertake wider horizon scanning to identify where genomics could be used as part of screening services to improve existing pathways for both sensitivity and/or specificity, and use this to evaluate when/if Wales would be able to implement these improvements *(PHGP)*

Population health genomics testing strategy

Genomics can be used in population health to either look for rare or other conditions as part of screening programmes or to predict when individuals are at a high relative risk of developing certain common diseases. The population health testing strategy will utilise inherited genomic information to direct clinical care through prevention and early detection of disease.

A range of genomic technologies are available to support targeted or region-specific genomic assays in cancer, rare disease and population health. Currently, genomics is used as part of screening programmes in Wales, for example through non-invasive prenatal testing for common aneuploidies. Current screening research programmes such as Our Future Health and The Generation Study will impact future healthcare screening diagnostic testing pathways to enable early clinical management or treatment interventions. Such screening programmes will be mandated by Welsh Government and facilitated through Public Health Wales.

ACTION 2.34: Identify individuals at high risk of adverse drug reactions and prepare for pre-emptive pharmacogenomic testing, delivered through panel testing (*Pharmacogenomics Delivery Plan for Wales*) *(AWMGS)*

ACTION 2.35: Deliver any changes required to the current provision of genomic testing for sudden infant death syndrome and sudden unexpected death in childhood resulting from the pathway review being undertaken by Welsh Government, NHS Performance and Improvement and Public Health Wales *(AWMGS)*

ACTION 2.36: Expand circulating biomarker testing as part of screening which will be driven by evidence *(AWMGS)*

ACTION 2.37: Evaluate potential legislative changes using population-wide genomics (including from initiatives like Our Future Health and The Generation Study); evaluate operational requirements to deliver testing, interpretation and reporting of e.g. polygenic risk scores and implementation of new services *(AWMGS)*

Pillar 4: Pathogen Genomics Services

Pathogen genomics involves examining microorganisms that cause diseases and using this information to inform or initiate action. Pathogen genomic data enhances and underpins health protection, by identifying and characterising outbreaks, and providing a basis for real-time pathogen surveillance. In addition, pathogen genomics also forms a cornerstone of precision medicine in deciding treatments for pathogens such as human immunodeficiency virus or tuberculosis. Pathogen genomics is well established in NHS Wales, routinely generating data that is utilised in real time at a range of scales, from individual patient to population. These accredited services are integrated into the wider Public Health Wales infection and health protection services, providing an All-Wales capability. The *Pathogen Genomics Delivery Plan for Wales* has been developed to outline the vision for building on the strength of this capability over the coming years.

Planned activities within the *Pathogen Genomics Delivery Plan* include improving existing services, developing new pathogen genomics services, and laying the groundwork for services that integrate human and pathogen genomics. There are also plans to integrate pharmacogenomics into public health surveillance systems to enhance population health outcomes, in alignment with the *Pharmacogenomics Delivery Plan*. Delivering the *Pathogen Genomics Delivery Plan* is the key action under Pillar 4, alongside a small number of other allied actions which reach beyond current areas of pathogen genomics activity in Wales.

ACTION 2.38: Implement the *Pathogen Genomics Delivery Plan* for Wales *(PHGP)*

In addition to the focused work detailed in the *Pathogen Genomics Delivery Plan*, we will also undertake activities to:

ACTION 2.39: Develop and implement an NHS Wales Pathogen Genomics Advisory Board *(PHGP)*

ACTION 2.40: Identify the core genomics-based toolset required to support the climate change surveillance system, based on a ONE Health approach, including vectors, hosts and animals as well as pathogens *(PHGP)*

ACTION 2.41: Integrate wider NHS and academic genomic capacity and capability planning into pandemic preparedness response plans *(PHGP)*

Pillar 5: Service Evaluation and Improvement

The Duty of Quality in healthcare | GOV.WALES outlines the NHS commitment to safe, effective and person-centred health services. Consolidation of existing services is a key focus for genomics partners to improve the care for patients. Service improvement will be achieved through several initiatives that will include better digitisation, evaluation and implementation of new technologies and methodologies, and increased collaboration. Embedding new genomic cancer applications and treatment diagnostic pathways will be integral to the vision of new models of care.

ACTION 2.42: Participate in the evaluation of the Rare Disease Lead Consultant Nurse role as part of the Rare Care Centre which aims to improve care for rare disease patients in Wales (*AWMGS*)

ACTION 2.43: Continue to provide specialist genomic expertise to multidisciplinary teams, health boards and trusts across the healthcare system *(AWMGS)*

ACTION 2.44: Implement appropriate patient questionnaires to capture Patient-Reported Outcome and Experience Measures to support continuous improvement for the genomics service (*AWMGS*)

ACTION 2.45: Develop an Electronic Patient Record for Clinical Genetics patients to support all-Wales working leading to reduced waiting times, improve access to patient data and clinical decision-making, and aid recruitment to clinical trials (*AWMGS*)

ACTION 2.46: Explore local implementation of functional genomics and other advanced genomic technologies to improve NHS diagnostic rates for rare disease *(AWMGS, WGP)*

ACTION 2.47: Enable discovery science using key techniques that include long-read sequencing and RNA *(AWMGS, WGP, PHGP)*

ACTION 2.48: Support the Syndrome Without a Name (SWAN) clinic, commissioned by the NHS Wales Joint Commissioning Committee, to provide greater capability to increase rare disease diagnostic rates, and access to precision medicines and clinical trials through reanalysis, including the development of a variant reanalysis service linked to NHS England's Test Directory *(AWMGS, WGP, PHGP)*

ACTION 2.49: Support Research and Development initiatives, with industry and other partners, including Cancer Vaccine Launchpad, long read sequencing and proteomics *(AWMGS, WGP, PHGP)*

ACTION 2.50: Undertake horizon scanning to identify where genomics could be used for potential new screening, population diagnostic or vaccination programmes. Potential areas include:

- The examination of microbial markers of cancer via a descriptive epidemiology study to establish if they could be used to improve specificity of bowel screening test
- Provide a less invasive method for certain types of screening activity, which could improve uptake by reducing barriers to attendance, especially with communities who might not otherwise engage
- Examine and evaluate genomic data to identify where vaccines may prevent chronic disease to support the provision of new vaccination programmes *(PHGP)*

ACTION 2.51: Evaluate the effectiveness of the cervical screening programme by exploring Human Papillomavirus to understand the impact of the vaccine programme on epidemiology of the virus *(PHGP)*

ACTION 2.52: Develop a methodological approach that integrates genomics and includes equity and social value in economic calculations into population health decision making *(PHGP)*

ACTION 2.53: Examine the impact of inequalities in sequencing access and sequencing data, and its potential impact on providing genomics-based healthcare and public health services in Wales *(PHGP)*

ACTION 2.54: Implement tools to interpret population-wide genomic results and other information to review the effectiveness of interventions *(PHGP)*

ACTION 2.55: Support academic and industry partners through provision of bioinformatic services and expertise, providing access to sequencing equipment and development of networking and regulatory infrastructure needed to enable genomic data sharing between partners *(WGP)*

Delivery Theme 3: Research

Wales continues to lead the way in advancing genomics, and our research expertise spans the full spectrum from discovering new genes and drug targets, through to implementation into patient services. Genomics is recognised as a key innovation in transforming healthcare; the full potential will only be realised by embedding genomics research and the translation of new opportunities into routine clinical care.

Increased access to clinical trials for patients in Wales will become an integral part of their care pathway. Equitable and consistent identification of eligibility for clinical trial participation will require new frameworks and systems to be developed. Investment through the Voluntary Scheme for Branded Medicines Pricing and Access (VPAG) programme will significantly expand commercial clinical trials capacity in the UK. In Wales, this will enable increased patient access to genomic-enabled clinical trials.

Wales Gene Park, All Wales Medical Genomics Service and Cardiff University School of Medicine are developing approaches using long-read and RNA sequencing with a view to transfer these into service delivery for patients within the next few years (Delivery Theme 2). Data innovation is being driven within the cancer and clinical neurosciences research communities, who are partnering with both internal and commercial experts looking at digital transformation and innovation. Much of this innovation is centred around exploiting information in many different data types (multi-modal data) including but not limited to genomic data.

As genomics continues to transition from research into routine healthcare it not only improves the patient experience but also accelerates the implementation of new services and fosters staff development. Research access to NHS genomic data enhances diagnostic rates, facilitates the identification of patients for clinical trials, and supports the discovery of novel disease associations.

Strengthening a cohesive genomics research landscape in Wales

There is a need for better co-ordination across genomics research in Wales. A commitment was made in the previous delivery plan to develop a genomics research strategy for Wales that would support research across areas of strength in Welsh higher education institutions, NHS organisations and other centres of excellence. *The Strategic Research Plan for Genomics in Wales* also outlines objectives to drive the ethical use of genomic data for research (including consent and governance frameworks), and interoperability of datasets across Wales, the UK and internationally. Moreover, the structure and scale of NHS Wales offer a strong foundation for close collaboration and rapid translation of internationally recognised research from Welsh academic institutions into clinical practice, but this potential is not yet fully realised. The ambition in Wales is to create a formal 'translational interface' to allow research insights to be returned to NHS Wales clinical teams for evaluation and implementation, detailing how research will directly inform the improvement of existing genomic services. Fostering stronger integration across organisations will be key to achieving this.

ACTION 3.1: Establish the required framework to support the translation of proven research into clinical practice and policy through partnership arrangements and collaborations across the NHS, academic and industry sectors *(WGP, PHGP, AWMGS)*

Although genomics leads healthcare research in both the scale of data generation and the potential for novel patient insights, it does not exist in isolation. The future frontiers of healthcare research lie

in integrating and mapping data from across the full spectrum of healthcare interactions and even extending them into all aspects of a patient's life. We must therefore seek to strengthen bonds with other cross-partner research organisations such as the Cardiff Cancer Research Partnership, the Wales Imaging Academy, the Secure Anonymised Information Linkage (SAIL) Databank team and the research teams within Digital Health and Care Wales. Many links exist between genomic researchers and these organisations at an operational level, but shared strategic aims and consensus is required for the future.

ACTION 3.2: Explore the opportunities to establish strategic mechanisms for collaboration with related research and healthcare partners (for example Cardiff Health Partners including Cardiff Cancer Research Partners, and Secure Anonymised Information Linkage (SAIL) Databank) to strengthen cross-organisational working where needed *(WGP)*

Partnering with patients

The Patient and Public Sounding Board has demonstrated the impact of involving patients to shape the genomics research strategy via their regular consultations (*see Delivery Theme 1*). It is key that we continue to use this established framework to build in patient expectations around data usage for research purposes.

ACTION 3.3: Use patient involvement frameworks and activity to ensure patient expectations are included in our longer-term vision that all NHS patients in Wales undergoing genomic testing will be offered the opportunity to consent to research access to their data (*WGP)*

Exploitation of broader capabilities for Welsh research

There is a need to exploit broader capabilities in genomics research and delivery across Wales with closer working between genomics partners and Health and Care Research Wales (as shown in the *Strategic Research Plan for Genomics in Wales*); there is also an opportunity to use the advantage of proximity of human and pathogen services at Canolfan Iechyd Genomig Cymru for potential future services

ACTON 3.4: Establish a methodology for genomics surveillance; support the rapid identification of emerging pathogens and changing distribution of variants for emergency preparedness *(PHGP)*

ACTION 3.5: Identify the options to utilise human genomic testing in population screening where individuals at risk are not identified by current services; this will require linkage across NHS services *(GPW)*

ACTION 3.6: Explore opportunities in human-pathogen interactions, human-therapy interactions and gene-environment-human interactions (*PHGP, AWMGS, WGP)*

Contribution to research initiatives outside of Wales

Exploitation of opportunities to link genomics expertise in Wales with UK initiatives is important, alongside potential for international collaborations (as detailed in the *Strategic Research Plan for Genomics in Wales*). Whilst Wales demonstrates clear UK-wide and even international leadership in numerous key fields of human and pathogen genomics, the frontiers of research are now happening within disease and population samples at a scale beyond what can be delivered with a purely

localised focus. For the purposes of discovery, there is a need to work with national and international partners to combine knowledge, samples and data to provide the resources needed for future genomics innovation.

ACTION 3.7: Share details of Wales's genomic datasets accessible via existing research gateways (for example All Wales Genomic Databank, Secure Anonymised Information Linkage (SAIL) Databank, Wales Cancer Biobank) by adding to national data-catalogues (such as the Health Data Research UK Health Data Research Gateway) allowing visibility of our resources and a route to forge new research collaborations *(WGP)*

Providing researchers with secure access to NHS genomic data linked to clinical records

Following the successful pilot stage, anonymised NHS genomic data will continue to be transferred to the Secure Anonymised Information Linkage (SAIL) Databank, which employs robust safeguards to prevent individual identification from healthcare and demographic data. As such, individual-level consent is not required for data submission to this databank.

The ambition is for genomic analysis reports generated by All Wales Medical Genomics Service to be shared with the Secure Anonymised Information Linkage (SAIL) Databank with data access guided by ethical approval, governance and researcher need. Raw and unprocessed, diagnostically active genomic data will be securely held within the All Wales Medical Genomics Service digital infrastructure. This will be a hybrid on-premises / cloud solution. The ambition is to enable legal access to All Wales Medical Genomics Service raw data through the creation of a secure data environment.

Whilst anonymised access is powerful at a population level, consent is vital to allow feedback of clinically significant findings, permit extraction of data on individuals or small cohorts (less than ten people) and allowing recontact of individuals for future research and clinical trial recruitment. It will be essential to track which individuals have provided consent for research access. Wales already has existing data infrastructure capable of supporting systematic and ongoing service monitoring and evaluation. However, for this to be meaningful, NHS genomic data must be routinely linked with clinical outcomes, prescribing information and other health records to assess the real-world impact of genomic interventions. The Wales Gene Park Data Integration team have established exemplar projects facilitating sharing of NHS genomic data in both cancer and rare disease cohorts using both consent and anonymised access models.

A key priority will be the development of secure data environments and systems that maintain patient privacy and anonymity while enabling advanced genomic analyses. Ideally, this would be through a federated network of specialised research environments with standardised Application Programming Interfaces and governance frameworks that promote collaboration across Wales and the wider UK.

ACTION 3.8: Guided by scientific priority and researcher needs, assess the volume and types of data generated by All Wales Medical Genomics Service that should be shared with the Secure Anonymised Information Linkage (SAIL) Databank *(WGP, AWMGS)*

ACTION 3.9: Collaborate with data controllers for large datasets relevant to multi-modal analysis (e.g. histopathology and medical imaging) to map out secure methods for data linkage that enable federated analysis workflows without data transfer, under appropriate governance *(WGP)*

ACTION 3.10: Develop governance frameworks and access mechanisms, in collaboration with relevant stakeholder (including academic researchers), to define how genomic data will integrate into the future landscape of multi-modal data for both service development and research purposes *(WGP)*

The digital strategy for genomics is under development and will provide details of the current research infrastructure and plans to meet the future requirements to support research ambitions.

Improving access to biological samples linked with clinical and genomic data for research purposes

The current frontier in utilising genomic data generated within NHS Wales for research, is to move beyond standalone biobanks or data repositories towards developing an integrated bioresource that combines both. This approach will enable researchers to identify and access specific biological samples for novel laboratory investigations. Wales's biobanks and bioresources are well positioned to support this transition, making available data from a patient's full healthcare journey alongside biological samples, for academic and commercial research.

All Wales Genomic Databank

We will formalise a 'single secure route' for NHS Wales generated genomic data to enable streamlined access for Welsh researchers (while ensuring data remains within Wales's secure governance frameworks) using the genomic research consent system, All Wales Genomic Databank. We will facilitate seamless and secure linkage with UK-wide genomic and health datasets (such as Our Future Health and UK Biobank). All Wales Genomic Databank has been designed to ensure interoperability with other Welsh biobanking initiatives, such as Wales Cancer Biobank and Cardiff University Biobank. This alignment will facilitate resource sharing and create opportunities for future integration, enhancing efficiency and scalability. Our long-term vision is that all NHS patients in Wales undergoing genomic testing will be offered the opportunity to consent to research access to their data, and to be recontacted for future research participation. We will work with our partners to develop All Wales Genomic Databank to become a fully featured bioresource which integrates the patient's healthcare journey as well as making biological samples available for academic and commercial research.

Long-term objectives (from 2028 onwards) will focus on maximising the utility of consented data and samples by increasing cohort size, improving access and achieving financial sustainability for the All Wales Genomic Databank initiative.

Resources are required to optimise and simplify existing methods and infrastructures that support linking NHS Wales clinical genomic data to biological material and making both available for research. Ideally, this should be via a 'single secure route' such as the All Wales Genomics Databank. Consent mechanisms will be central to this approach, allowing data and associated biological material to be used responsibly while ensuring clinically relevant findings can be fed back to patients.

ACTION 3.11: Enable efficient feedback of clinically relevant findings to patients from research through All Wales Genomics Databank framework *(WGP)*

ACTION 3.12: Work with partners to develop the All Wales Genomic Databank into an integrated bioresource that spans the patient's healthcare journey and provides multimodal healthcare data available for academic and commercial research *(WGP)*

ACTION 3.13: Collaborate with biobanks and All Wales Medical Genomics Service to plan for the accompanying expansion in data scale, systems and workforce capacity *(WGP)*

ACTION 3.14: Maximise the value of consented data and samples by expanding cohort size, improving access, and ensuring the financial sustainability of Welsh bioresources through research use *(WGP)*

Delivery Theme 4: Enablers

Digital and Data

The storage, processing and use of genomic data creates a growing challenge for current digital infrastructures; robust planning for digital systems, connectivity, security and information governance is required to support the wider service and research ambitions for genomics.

The digital strategy for genomics is under development. This will detail the current digital environments across the partners, the plans for changes to support genomic services and digital requirements to meet current and future needs from both a service and research perspective; this will include digitisation and automation of genomic laboratory test requesting and reporting and interoperability with wider NHS systems such as NHS Wales App.

Appropriate governance structures need to be established to support the sharing of digital environments and genomic data by multiple organisations.

ACTION 4.1: Publish a digital strategic roadmap for genomics that describes how the requirements of service and research partners will be fulfilled *(GPW, DHCW)*

ACTION 4.2: Establish appropriate policies, procedures and contracts that meet legal information governance, cyber security and data protection requirements, and facilitates the appropriate and effective sharing of equipment and data for agreed purposes (including clinical delivery, population health and research) *(GPW, AWMGS, WGP, PHGP)*

ACTION 4.3: Ensure industry and academic partners in Wales have access to bioinformatic and genomic data analysis expertise through provision of appropriate services *(WGP)*

ACTION 4.4: Store All Wales Medical Genomics Service data in a Cloud environment as part of a hybrid data storage strategy *(AWMGS)*

ACTION 4.5: Make All Wales Medical Genomics Service reports generated by the Genomic Laboratory Information Management System interoperable with Digital Health and Care Wales clinical systems (*GPW, AWMGS, DHCW*)

ACTION 4.6: Develop a Secure Data Environment for cloud based All Wales Medical Genomics Service data to enable access *(AWMGS)*

Infrastructure

Premises – the co-location of Genomics Partnership Wales partners at Canolfan Iechyd Genomig Cymru will see several benefits realised. Within the new facility, there are physical enabling areas including accredited laboratories, research laboratories, collaboration spaces, modern patient clinics and seminar rooms appropriate for public and professional engagement.

ACTION 4.7: Evaluate with all partners at Canolfan Iechyd Genomig Cymru the current and ongoing performance against the investment objectives outlined in the Genomics Partnership Wales Benefits Realisation Plan, and capture any new transitional or long-term benefits that were not included in the original plan *(GPW)*

Equipment – genomics research and services require a wide range of equipment and there has been significant investment over the last few years to support new technologies and the increase in demand particularly for genomic testing.

ACTION 4.8: Maintain a capital requirements list for the Partnership, including costs, priorities and impact to allow the utilisation of funding opportunities as they arise *(GPW)*

ACTION 4.9: Develop a strategy for equipment use at Canolfan Iechyd Genomig Cymru to ensure efficiency and to demonstrate the benefit of co-location *(GPW)*

ACTION 4.10: Ensure industry and academic partners around Wales can access genomic sequencing equipment; this will require development of networking infrastructure and regulatory approvals to allow transfer of genomic data *(WGP)*

National clinical and diagnostic pathways

Equitable access for patients to timely diagnoses and personalised medicines across Wales requires a system-wide approach to implementation of services. The clinical delivery of new services and treatments, particularly complex therapies, may require changes to the patient pathway such as increased capacity in existing areas or new methods. Horizon scanning and planning for system preparedness is required to deliver new treatments to patients.

ACTION 4.11: Enable closer collaboration with the National Pathology Programme to ensure adoption and embedding of genomics into the cancer clinical pathways; improve efficiencies and turnaround times across the whole diagnostic testing pathway, ensure fixation processes are standardised for optimal nucleic acid extraction and avoid duplicate testing between the services *(AWMGS)*

ACTION 4.12: Play an active role in the national medicines' optimisation approach; this includes the national pathways mapping of complex/novel therapies into practice and Systemic Anti-Cancer Therapies Horizon Scanning Collaborative Group *(AWMGS)*

ACTION 4.13: Work with colleagues across the UK to develop guidelines to support the transition to routine use of metagenomic sequencing to identify microbial infections *(PHGP)*

ACTION 4.14: Establish a framework to support the development and adoption of advances in microbial therapies, including faecal microbial transplantation, utilising genomic approaches *(PHGP)*

ACTION 4.15: Work with the Healthcare Associated Infection, Antimicrobial Resistance and Prescribing Programme to explore and, where appropriate, implement opportunities to integrate genomics into antimicrobial resistance surveillance and stewardship *(PHGP)*

ACTION 4.16: Collaborate with the Vaccine Preventable Disease Programme to explore opportunities in the application of genomics in the development of new vaccines and in measuring vaccine effectiveness *(PHGP)*

Commercial Partnerships: Innovation Ecosystem

In addition to improving healthcare, genomics provides potential benefits to the economy in Wales as part of the wider life science industry. Over the past three years, Genomics Partnership Wales has worked to establish the co-location of partners at Canolfan Iechyd Genomig Cymru, the genomics

centre of excellence for Wales. The vision behind the co-location was not just to foster collaboration and bring together genomics activity; it was driven by the desire to build on the world leading activities within the Welsh NHS to support the development of the wider life sciences ecosystem in Wales. The next phase of development for Genomics Partnership Wales must prioritise commercial partnership and ecosystem development, using our facilities and capabilities to sell Wales to the world. Working closely with Welsh Government and the Capital Regions, there is an opportunity to work as a partnership to attract inward investment and create new high value jobs in the life science industry clusters in Wales.

Our ambition

Our ambition is to capitalise on the strategic co-location of partners at Canolfan Iechyd Genomig Cymru to position Wales as a UK and global leader in harnessing genomics expertise to drive economic growth. We plan to do this by aligning public sector innovation with private sector investment, workforce development and UK industrial strategy priorities through the Genomics Partnership Wales programme. To achieve this, Wales needs to be outward looking and ready for business.

The *Genomics Delivery Plan for Wales 2022–2025* outlined a clear and ambitious approach to commercial partnerships as a key enabler of its vision. The plan emphasised that commercial partnerships are central to Wales becoming an international hub for genomics and precision medicine to drive better health outcomes, attract global commercial partners and create jobs and economic growth.

There has been good progress in this area. The co-location of partners to Canolfan Iechyd Genomig Cymru has provided a focal point and 'front door' for genomics and life sciences in Wales and has attracted interest from international trade delegations and life sciences organisations. It is part of an ambitious prospectus for the Cardiff Edge site which creates physical and digital links with the new Velindre Cancer Centre at the north end of Cardiff and continues along a five-mile innovation corridor. It connects key resources and infrastructure in the form of Cardiff University's campuses and NHS sites, including University Hospital of Wales, into a single translational ecosystem, co-locating discovery science, clinical trials, advanced diagnostics, digital health and patient care.

The Cardiff Edge site has strategic links along the A470/M4 corridor to the indigenous Life Science Cluster and into West Wales connecting clinical assets and higher education institutions such as Swansea University, Swansea Bay and Hywel Dda University Health Boards, and eastwards across the GW4 Alliance (a strategic alliance between Cardiff, Bristol, Bath and Exeter Universities).

Genomics Partnership Wales partners have engaged with numerous initiatives and stakeholders to date, including Life Sciences Hub, Cardiff Health Partners, health board and trust innovation teams, universities and the Wales Cancer Industry Forum, and has delivered closer working relationships with Illumina (through a Memorandum of Understanding), and Welsh Government trade and industry teams. Building on these collective activities, and the existing relationships that each partner also possesses, there exists a strong foundation for commercial partnerships focused on industry collaboration, data-driven innovation and public-private partnerships. In turn, these activities will incubate the infrastructure, data and innovation ecosystems that are essential for commercialisation.

ACTION 4.17: Explore the opportunities across Wales by engaging with areas of expertise related to genomics and life sciences to leverage capabilities and enable innovation activity in the wider Welsh ecosystem *(LSHW)*

Importance of genomics in the UK

The *UK's Modern Industrial Strategy: Life Sciences Sector Plan* aims to create a national genomic knowledge base that will provide real-world data to inform academic and industry research and development. Wales is recognised in this plan as one of only three UK regions to have commercially attractive strengths in genomics. This is a glowing endorsement of the work already undertaken in this space.

An action included in the *Life Sciences Sector Plan* is to 'Significantly expand commercial clinical trials capacity via funding from the Voluntary Scheme for Branded Medicines Pricing and Access (VPAG) programme'. This investment programme is an agreement between the Department of Health and Social Care on behalf of all nations of the UK and the Association of the British Pharmaceutical Industry, which is the representative body on behalf of branded medicine pharmaceutical industry in the UK. The purpose of the clinical trials aspects of the programme is to build dedicated capacity and capability for commercial research and therefore plays a role in enabling significant inward investment that will improve clinical trials in the UK. One aspect of Wales's involvement in this programme is to enable increased patient access to genomic-enabled clinical trials.

The *NHS England 10-Year Plan* places genomics at the heart of the future of healthcare. This vision is further detailed in the *Accelerating Genomic Medicine in the NHS*, which outlines how genomics will be embedded across the NHS in England over the next five years. NHS England aims to create a national genomic knowledge base that will provide real-world data to inform academic and industry research and development. This data infrastructure is designed to support innovation pipelines, enabling companies to develop new diagnostics, therapies and digital tools. Genomic data will be integrated with other clinical and diagnostic data to support Artificial Intelligence-driven insights, clinical decision support and population health management.

In response to this, Genomics Partnership Wales needs to cement its position as a platform for innovation, enabling companies to develop, test and scale genomic solutions within a real-world healthcare setting. Genomics Partnership Wales will need to be agile and responsive to the UK policy drivers, be involved in UK initiatives that are of benefit to the population of Wales and identify opportunities where Wales has a strength or something unique to offer.

Genomics Partnership Wales will work with partners like Health and Care Research Wales, Office for Life Sciences, Genomics England, UK Research and Innovation, and industry stakeholders to accelerate the translation of genomic science into clinical practice, to drive efficiencies, support innovation and enable commercial applications of genomic technologies for the benefit of patients and economic growth in Wales.

Any Welsh multimodal data assets must be developed using common standards ensuring interoperability with other UK assets but still be seen as attractive to commercial partners on a

national level in Wales particularly regarding the Secure Anonymised Information Linkage (SAIL) Databank and Digital Health and Care Wales's National Data Repository.

For the next phase, Genomics Partnership Wales will look to maximise the potential of existing investment in genomic infrastructure over the next three years.

ACTION 4.18: Organise an annual innovation showcase event to engage commercial and academic partners and potential investors *(GPW, LSHW)*

ACTION 4.19: Develop a sector focused marketing plan, associated skill set and commercial offer to support attendance at trade exhibitions with a strong commercial partnership focus *(GPW, LSHW)*

ACTION 4.20: Develop a legal and ethical commercialisation model for genomic processed and raw data to facilitate working with commercial partners *(AWMGS, PHGP)*

ACTION 4.21: Consider the contribution of genomic data to a multimodal national data asset (at either Welsh or UK level) which will be interoperable with other data assets permitting the widespread deployment of Artificial Intelligence tools. This will involve engagement with Health Data Research Service to determine the opportunity for data platform and infrastructure readiness for scale subject to Welsh Government policy *(WG SRE)*

ACTION 4.22: Align this delivery plan with the UK Industrial Strategy's sectors (particularly Life Sciences, Digital and Technologies, and Professional Services) to ensure we can participate in the relevant UK-wide activities, and identify any opportunities for Wales to offer something unique *(LSHW)*

ACTION 4.23: Embed genomic data, research and services into NHS pathways via pilots and commercial-public delivery models *(GPW, LSHW)*

ACTION 4.24: Complete genomics sectoral analysis in the context of the UK's Modern Industrial Strategy particularly the Life Sciences Sector Plan *(LSHW)*

ACTION 4.25: Develop a framework to assess the economic impact across Wales (jobs, inward investment, revenue, co-licensing opportunities) to allow the evidence to be collated *(LSHW)*

ACTION 4.26: Strengthen links with Wales Cancer Industry Forum; explore running scoping workshops to identify high-impact collaboration areas, for example pharmacogenomics, liquid biopsy, pathogen sequencing *(LSHW)*

ACTION 4.27: Evaluate and plan to develop a rare disease commercial partnership forum with Rare Disease Implementation Network, including NHS, academia, industry representatives and patient advocates *(GPW)*

ACTION 4.28: Work with Advanced Therapies Wales to create a national precision medicine commercial partnership offer *(GPW)*

ACTION 4.29: Agree programme of works with Genomics England aligned to the *Life Sciences Sector Plan (GPW)*

ACTION 4.30: Develop an ambitious programme of work to attract global diagnostics, pharmaceutical, biotechnology and medical technology investors. Areas for exploration include a Wales-wide investment summit, a national genomics innovation accelerator, a national genomics commercialisation hub, and embedding commercial teams at the Cardiff Edge site to create a community for engagement and collaboration between industry, academia and the NHS *(GPW)*

Planning, Funding and Commissioning

The future funding model for all the genomics services and programme activities needs to be considered (see *Long Term Sustainability* below). There is a need to ensure that genomics is included appropriately in the planning processes for NHS organisations on an annual basis to provide focus and priority.

ACTION 4.31: Include genomics in the appropriate plans (Integrated Medium-Term Plans, Clinical Service Plans) for all NHS Health Boards and Trusts to reflect the activity needed to support mainstreaming of genomics within each organisation and/or support genomics delivery *(GPW)*

ACTION 4.32: Include genomics awareness and education in the workforce plans for all NHS Health Boards and Trusts *(GPW)*

ACTION 4.33: Review the commissioning model for the pathogen genomics services *(GPW, PHGP)*

Horizon Scanning

There are several organisations in Wales that perform horizon scanning, and these can be in medicines and therapies, technology, and clinical relevance and understanding. Genomics Partnership Wales will establish a systematic process of identifying, analysing and evaluating emerging trends, technologies, research and innovations in the field of genomics that may have significant implications for healthcare, public health, policy and service delivery in the future. This involves monitoring scientific advances and assessing their potential impacts on clinical practice, diagnostics and population health. Ethical, legal and social implications will need to be considered, and inform strategic planning and investment decisions made within the genomics programme.

ACTION 4.34: Establish a framework for genomics horizon scanning that aligns to our strategic priorities. Utilise information produced by organisations who undertake horizon scanning activities to identify opportunities, work with subject matter experts to assess potential impact and use this intelligence to inform Genomics Partnership Wales strategic risk management *(GPW)*

Environmental sustainability

Reducing the carbon footprint of the genomics activities is a key area of focus.

ACTION 4.35: Work collaboratively with genomics partners and other Diagnostics programmes to establish a mechanism to monitor and report on environmental sustainability measures put in place across the genomics ecosystem *(GPW)*

ACTION 4.36: Evaluate and implement improvements in service to reduce our carbon footprint as part of our sequencing and analysis *activities (AWMGS, PHGP, WGP)*

Delivery Theme 5: Strategic Programme Functions

To ensure a collaborative, strategic approach to genomics in Wales, the Genomics Partnership Wales national programme will drive the delivery of this Plan ensuring adherence to appropriate governance arrangements and regular monitoring and reporting of progress.

Governance

The governance of the Genomics Partnership Wales programme is complex, reflecting the different organisations involved and the different areas of focus. Diagnostics elements are overseen by the Planned Care Board, and for some areas there is direct accountability to Welsh Government. Each key partner has accountability to their hosting organisation.

Evaluation

Demonstrating the value of the programme and genomics service implementations to patients, the healthcare system and the wider economy is key. We will take the opportunity to use the Patient and Public Sounding Board to hold the programme to account for deliverables. We will also work with any established expertise in Wales to demonstrate the value of genomics for patients and the wider system.

ACTION 5.1: Develop a framework and dashboard that identifies, defines and captures benefits from all workstreams within the Genomics Partnership Wales programme ensuring alignment to the intended impact or value *(GPW)*

Long term sustainability

The sustainability of the genomics programme and Wales Gene Park (who are both currently on short term budget arrangements) needs to be resolved to provide stability to the genomics ambitions and plans.

Advanced Therapies Wales is closely aligned to Genomics Partnership Wales in several workstreams and there are efficiencies to be gained from working together in these areas.

ACTION 5.2: Explore the options for a long-term funding model for Genomics Partnership Wales and Wales Gene Park *(GPW)*

ACTION 5.3: Work more closely with Advanced Therapies Wales in identified areas that align to demonstrate efficiencies, increased value and mutual benefit *(GPW)*