A Strategic Research Plan for Genomics in Wales

Ymchwil Iechyd a Gofal Cymru
Health and Care Research Wales

Partneriaeth Genomeg Cymru
Genomics Partnership Wales

Ariennir gan **Lywodraeth Cymru**
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Foreword

Jeremy Miles MS, Cabinet Secretary for Health and Social Care

Wales continues to lead the way in advancing genomics, with initiatives like the newly established Canolfan Iechyd Genomig Cymru at Cardiff Edge exemplifying its transformative potential in health and care. The rapid evolution of genomics offers deeper insights into disease, improved clinical outcomes, and the ability to positively impact countless lives as well as driving significant opportunities for economic growth.

Welsh university research is driving real benefits for communities across Wales, the UK, and globally. Our expertise spans the full translational continuum: from discovering new genes and drug targets, to shaping diagnostics and treatment design, conducting clinical trials of personalised medicines, and implementing findings into frontline healthcare services.

This swift progress in genomics research must increasingly be embedded within core medical specialities to unlock its full impact. In developing this plan, we've highlighted Wales' strengths and the strategic objectives now seek to reinforce these domains, while nurturing vital emerging fields such as pharmacogenomics and functional genomics.

The Genomics Delivery Plan made a clear commitment to develop a dedicated research strategy for genomics. It's inspiring to see the Welsh research community come together to make that vision a reality. I'm eager to see the progress ahead and how it will fuel world-class research, grow a skilled and sustainable workforce, and lead to better health outcomes for the people of Wales.

Welsh Government Professor Isabel Oliver, Chief Medical Officer

Genomics provides us a deeper understanding of how our genes influence disease, treatment response, and overall health. Advances in genomic technology are resulting in more rapid and accurate diagnosis, new effective treatments and vaccines against important public health threats. Genomic research is important in our efforts to secure a healthier Wales. This research improves our understanding of diseases, enables development of diagnostics tests and help us develop new treatments. These rapidly evolving areas of research can also help identify those at risk of certain conditions such as a rare disease or cancer and are therefore key in prevention and early intervention for many serious conditions.

This research plan builds on Welsh research excellence and expertise in genomics research. It will help to ensure

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these strengths are reflected in national and UK implementation plans so that Welsh researchers are able to lead and contribute to the opportunities arising from the UK-level Shared Commitments for better health and wellbeing.

Genomics Partnership Wales – Suzanne Rankin, Chief Executive Officer Cardiff and Vale University Health Board and Senior Responsible Officer for Genomics Partnership Wales

At Genomics Partnership Wales, we are committed to building an internationally competitive genomics research environment. We are doing this through strategic investment in advanced genomic technologies and precision medicine platforms, the development of collaborative infrastructures including our patients, close coordination with partners such as Advanced Therapies Wales, and the delivery of an ambitious training portfolio as part of the Genomics Workforce Plan.

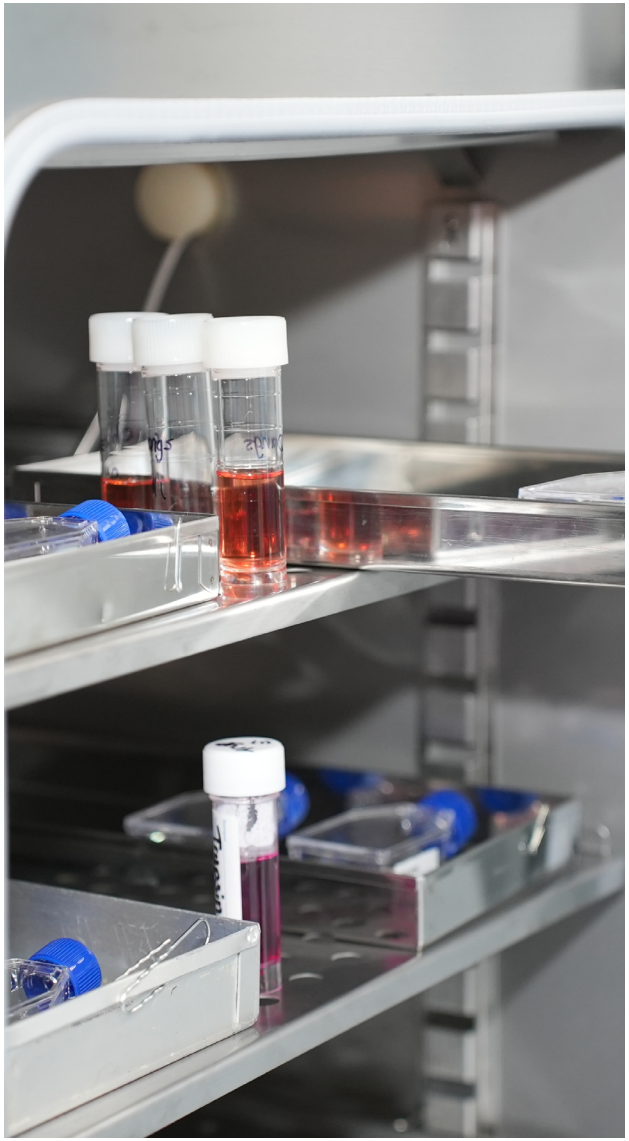
Links with industry will be more important than ever to maximise the success of our plans and to create health and economic benefits for Wales. This strategy will enable us to establish Wales as an outward-looking, collaborative and reputable home for business development, promote our genomic services, and attract the best partnership opportunities

Genomics Partnership Wales Patient and Public Sounding Board

Genomics research holds incredible potential, but patients need assurance that this work will translate into real, positive change for the lives of themselves and their loved ones. They want to see a commitment to inclusivity, making sure diverse populations are represented so the positive outcomes apply to all, not just a few.
While it is recognised that the possibilities are exciting, it is important to be transparent about what is realistic, both in terms of timelines and risks. Above all, privacy and trust are paramount and as genomic data is deeply personal, guarantees are needed about how it will be protected and used responsibly.

This plan promotes open communication between researchers and patients, aligns efforts with healthcare providers for smoother implementation, and aims to provide ethical, impactful outcomes. Members of the Sounding Board welcome their role in shaping this plan and look forward to ongoing involvement in its execution.

Summary



The Genomics Delivery Plan for Wales 2022-25 describes how Genomics Partnership Wales works with Welsh Government and other stakeholders, to harness advances in the understanding and application of genomics to transform public health strategy and delivery of care.

Within the plan, a commitment was made 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.

Informed through advice from an external advisory group (Annex one), a review of genomics research in Wales, and a joint Health and Care Research Wales and Genomics Partnership Wales community wide engagement exercise, this document 'A Strategic Research Plan for Genomics in Wales' 2026-2030 aims to highlight the scientific capabilities and research strengths we have in Wales. It also recognises the critical role that developments in workforce, data and infrastructure play in advancing our genomics ecosystem, leading to improvements for patients and economic benefits such as cost savings and generation of jobs.

The Strategic Research Plan sets out four objectives and the associated actions that will be taken forward over the next five years. The implementation of the plan will enable the Welsh research community to build upon existing areas of expertise. This will accelerate advances in genomic research, making new discoveries about diseases and improving healthcare and outcomes for the population in Wales and beyond.

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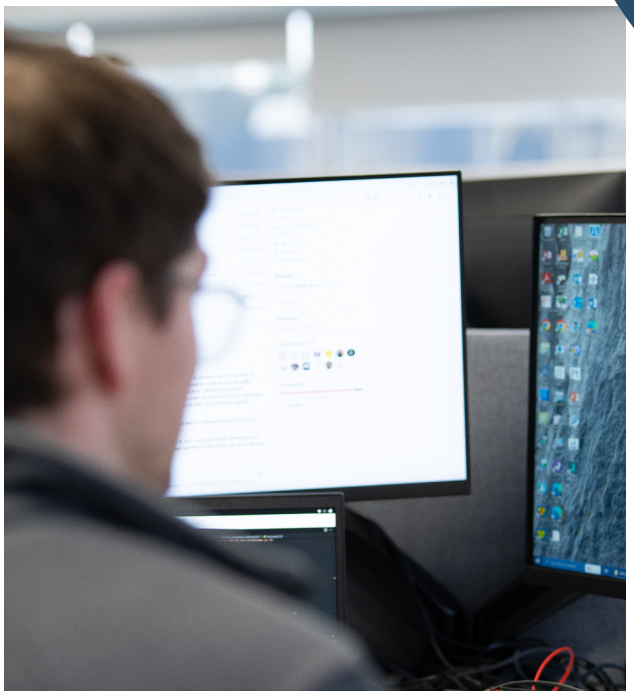
UK and Welsh Context

In 2017, the Welsh Government published the Genomics for Precision Medicine Strategy. This Strategy set out the Welsh Government's plan to create a sustainable, internationally competitive environment for genetics and genomics to improve health and healthcare provision for Wales and resulted in the establishment of Genomics Partnership Wales in 2018.

In 2020, Wales signed as a partner the UK Government's ten-year strategy Genome UK: The Future of Healthcare with the ambition for the UK to have a world-leading genomic healthcare system for the benefit of patients. In 2022, Genome UK: Shared Commitments for UK-wide implementation 2022 to 2025 was published to describe how the four nations would work together during this period. The Genomics Delivery Plan for Wales was developed to describe the Welsh implementation of Genome UK, which aligns with the Diagnostics Recovery and Transformation Strategy for Wales (2022-25). Also in 2022, NHS England produced their document Accelerating genomic medicine in the NHS. A strategy for embedding genomics in the NHS over the next 5 years. This was followed in 2023 by Genomics England publishing their research vision, Genomics-enabled research to drive healthcare for the benefit of everyone, with a focus on cancer and rare diseases and outlined three main challenges in ethics, AI, and data storage and computational power.

In parallel to this, Welsh Government has put in place multiple policies looking to drive innovation and transformation in health and social care. Some of those have specific relevance to genomics research and include A Healthier Wales, Digital Strategy for Wales, Advanced Therapies Wales Delivery Plan, Innovation Strategy for Wales and CReSt, the Cancer Research Strategy Wales. One of the key activities within the implementation of CReST - precision and mechanistic oncology, examines how genetics influences who gets cancer, how that



cancer behaves, and finding ways to treat cancers with certain genetic signatures.

This work is set in the broader context of the UK Government's Industrial Strategy, the Life Sciences Sector Plan and the NHS 10 year plan, that together aim to improve the health and wellbeing of citizens by driving economic growth and

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promoting the UK as a world-leading destination for life sciences companies. This includes the development of strategic partnerships with Moderna and BioNTech around their mRNA vaccine trials during 2024, of which Wales developed bespoke strategic agreements with each company internationally, countries such as Scotland, Denmark, Sweden and Finland are setting benchmarks by routinely linking genomic and health data to foster rich research ecosystems. Wales is actively advancing its own leadership in this space, drawing on insights from these exemplars to shape a distinctive, population-focused approach to genomic innovation and healthcare integration.

Additionally, Lord O'Shaughnessy's review of commercial clinical trials in 2023 and the four nations' response highlighted the importance of seizing opportunities of using genomics data to accelerate clinical trials. The investment into Wales via the Voluntary scheme for branded medicines Pricing, Access and Growth (2023) provides opportunities to strengthen Wales' capability to deliver genomics driven commercial trials. Wales' development of a One Wales approach to delivering research across NHS Wales and Wales-wide access for patients into a single research site, also provides a strong foundation for taking these trials forward at pace and increasing our portfolio of trials, particularly in areas such as advanced therapies, neurodegenerative diseases, cancer and rare diseases.

Development of the Strategic Research Plan for Genomics in Wales

Genomics is a golden thread that runs through several areas of active research where Wales has established strengths. These include mental health, dementia, cancer, rare disease, and pathogen genomics. More recently, Welsh researchers have been establishing reputations in new areas of genomics activity such as liquid biopsy, long-read sequencing, pharmacogenomics, functional genomics and wastewater-based epidemiology to track pathogens at a population level. Collectively, these established strengths and new developments are underpinned by partnerships between Welsh Government, Welsh Universities, NHS Wales Health Boards and Trusts, and Industry, and position Wales as a potential leader in the field.

The Welsh Government's investments in genomics signifies a dedicated commitment to the integration of innovations in genomics into healthcare delivery, all of which in turn links to genomics research, complements CReSt and aligns with the priorities of the Advanced Therapies Wales programme and the Wales Rare Diseases Action Plan.

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Genomics Research In Wales Now

Recent reviews of the genomics research landscape in Wales alongside a landscape report commissioned by Genomics Partnership Wales and Welsh Government and prepared by IQVIA, identified priority research themes and cross cutting areas of strength. In addition, they have highlighted key capabilities and enablers for genomics research.

Priority Research Themes

Neuropsychiatric Genomics

Neuropsychiatric genomics research at Cardiff University's Centre for Neuropsychiatric Genetics and Genomics (CNGG) is home to over 150 researchers and more than 30 principal investigators. The CNGG has attracted over £115 million of investment in the last five years from Medical Research Council (MRC), Wellcome, European Union and United States National Institute of Health (US NIH) programmes, as well as from charitable and commercial organisations. The environment within CNGG (rated as four star in REF2021) provides facilities, training and collaborative opportunities for their teams of researchers across career stages. CNGG occupies the second and third floors of the Hadyn Ellis Building, a £30 million research facility and flagship building at Cardiff University's innovation campus. CNGG has been a world-leader in psychiatric genomics for the last decade and has led to many fundamental genomic discoveries through studies of both common and rare genetic causes in schizophrenia, bipolar disorder and neurodevelopmental conditions.

Dementia

The UK-wide Dementia Research Institute is a leading collaborative research effort with seven centres across the UK. Research at the Cardiff centre has made significant advances in unravelling the complexities of neurodegenerative disease, attracting over £70 million in research funds, and developing 12 Research Fellows, eight Group Leaders and four Emerging Leaders. It has also trained nearly 60 PhD students, to invest in the future-leaders in dementia research. Over the last seven years, the team in Cardiff have discovered around 100 genes associated with Alzheimer's risk or earlier onset in Huntington's disease. They have used genomics to identify those at the highest genetic risk of developing neurodegenerative diseases, implicating specific cell types in disease development, and identifying delivery a role for Alzheimer's disease risk genes in modifying Parkinson's disease. They have used genetics to uncover new disease mechanisms, such

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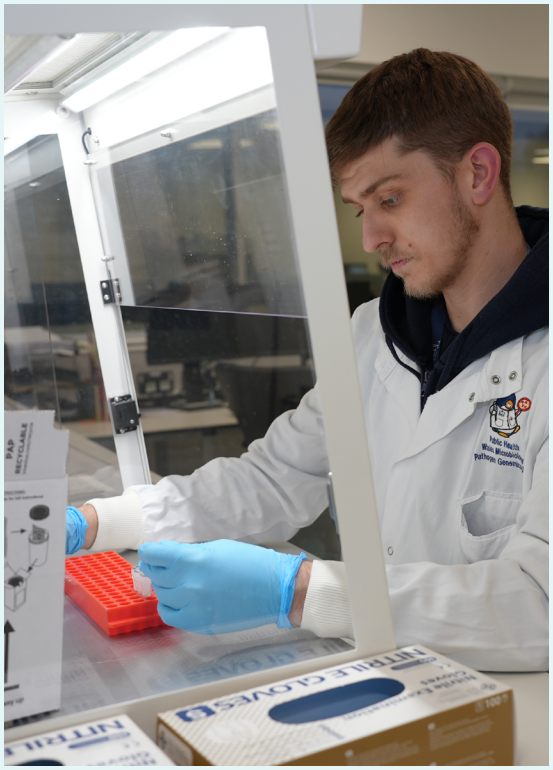
as microglial effects on synapses, endocytosis in glial cells, endothelial susceptibility and the complement system in Alzheimer's disease, and RNA repair in Huntington's disease. Looking to the future, activities in the Pentre Awel development in Llanelli will enable further medical research including the potential for dementia related genomic research and health care delivery, whilst supporting and encouraging people to lead active and healthy lives.

Pathogen Genomics

Within Wales, pathogen genomics activity has brought in more than £70 million in research income and is undertaken across a wide range of areas including antimicrobial resistance (including drug discovery); genomic epidemiology; microbiome and metagenomics; ONE Health, and infection biology/ microbial pathogenesis. Wales has a centralised microbiological laboratory network within the NHS with established strengths in clinical and public health pathogen genomics. Collaboration between this network and the Higher Education Institutions (HEIs) in



Wales is extensive, enabling the translation of research into practice e.g. C. difficile Whole Genome Sequencing characterisation and outbreak support. This was the first accredited national service of its type in the world. Welsh researchers played a major role in the planning of the COVID-19 Genomics UK Consortium (COG-UK), the development of the analysis platform for SARS-CoV-2, and the development of a genomic analysis pipeline and variant typer which are among the most frequently used in the world. Welsh researchers also led the development and design of MRC CLIMB (Cloud Infrastructure for Microbial Bioinformatics), the digital infrastructure upon which CLIMB COVID - the COVID sequencing analysis platform - was built. In addition, wastewater-based genomics led from Bangor University complements existing pathogen genomics efforts to support early warning systems for public health.

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Cancer

In Cardiff University, the Department of Cancer Genetics is developing research activity in prostate and bowel cancers and rare genetic diseases, with a particular focus on DNA damage and repair. Innovations like the INDUCE-seq® tool from their spin-out, Broken String Biosciences, spans mutation detection, genome-wide association, and next-generation sequencing analyses. Cardiff University and the NHS are collaborating in generating models of adoption for new technologies, specifically long read and RNA sequencing, in defined areas of strategic importance, including solid tumours (liquid biopsy samples) and haematological cancers. Strengthening the focus on longitudinal monitoring of tumour evolution using circulating tumour DNA (ctDNA) and real-time tracking of treatment resistance will ensure Wales remains at the forefront of precision oncology research.

Researchers at Cardiff University are actively developing bioinformatics tools to detect structural variants and visualise genomic data. These tools enhance the ability to interpret and communicate genomic data, facilitating more accurate and efficient research outcomes. In addition, Cardiff University has a significant long-term programme investigating telomere-driven genome instability in cancer. This research has led to the development of diagnostic, prognostic, and predictive tests that are commercially available to patients worldwide. These tests provide crucial information about disease progression and treatment response, improving patient care and enabling personalised medicine. Cardiff's Experimental Cancer Medicine Centre (ECMC) is actively involved in several innovative molecular trials, particularly in haematological malignancies and solid tumours and Velindre

University NHS Trust is a key stakeholder and enabler in this area providing essential expertise and assistance for genomic sequencing, patient involvement and the management and completion of clinical trials.

In North Wales, histopathology is

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spearheading efforts to enhance molecular pathology diagnostics, specifically in digital pathology, backed by collaborative projects aimed at establishing a Wales-wide, accessible near-patient testing framework, underpinning the growing focus on cancer and rare diseases as key research opportunities.

In Swansea University cancer research includes the identification of genetic, transcriptomic and epigenetic markers for endometrial cancers, ovarian cancers, colorectal cancers and oesophageal adenocarcinoma, and the development of analytical technologies for detection of epigenetic cancer markers (nanopore sequencing, and nanosensors for chromatin analysis). The implementation of ML/AI approaches to integrate multi-dimensional cancer data (imaging and omics) has led to the identification of new therapeutic targets, and the utilisation of Genomics England 100k genome data resources including the Cancer 2.0 longread data continues to expand.

Liquid biopsies, a priority in the Genomics Delivery Plan 2022–2025, are opening new paths for translational research in Wales. The All-Wales QuicDNA study, a collaboration between the NHS, Cardiff University, industry, the third sector, and patients, has integrated liquid biopsy technologies into lung cancer diagnostics, while enabling research into long-read sequencing of circulating tumour DNA (ctDNA). This work, supported by the All Wales Medical Genomics Service, has led to the QuicDNA Max programme, which will expand research to other tumour types and disease monitoring. Liquid biopsy research is advancing the use of genomic biomarkers for targeted therapies and clinical trial stratification. High-quality clinical and genomic data collection, supported by Digital Health and Care Wales, is critical. QuicDNA Max will also explore improved access to anonymised data via SAIL databank, including for conditions like clonal haematopoiesis of indeterminate potential (CHIP). These efforts further highlight Wales' growing leadership in real-world genomic research and precision oncology.

Rare Inherited Diseases Research

A rare disease is one that affects fewer than one in 2000 people, and 175,000 people in Wales will be affected by a rare disease at some point in their lives. There are over 6,000 rare diseases, more than 70% of which are genetic in origin. Welsh genomics research strengths are implicit across the portfolio of research described above e.g.

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through the rare genetic studies of schizophrenia, bipolar disorder and neurodevelopmental disorders such as Timothy syndrome and DiGeorge syndrome, rare forms of late onset Alzheimer's Disease and rare cancers such as the inherited tumour syndromes familial polyposis and Tuberous Sclerosis.

Work carried out in Swansea University and Cardiff University showed for the first time that mTOR inhibitors are an effective treatment for Tuberous Sclerosis Complex (TSC) leading to them becoming central to the treatment of this condition.

Clinical Genetics services are provided by an All Wales Medical Genomics Service team of Clinical Geneticists, Genetic Counsellors, Family History Coordinators and administrators. The team deliver specialist genetic services to individuals and families with, or concerned about, rare and/or inherited genetic conditions. They are a major driver of rare disease research, with highly trained cohort of physicians and genetic counsellors who are currently in the process of developing a dedicated research strategy focused on rare diseases within clinical services. This strategy will be closely aligned with the broader Genomics Research Plan.

A recent award from Wales Innovation Network has supported partners in Cardiff University, Swansea University, University of South Wales, Bangor University, Welsh Government, Life Sciences Hub Wales, Wales Gene Park and the Rare Diseases Implementation Network to unite scientists, healthcare providers, and clinicians to advance research, improve patient outcomes, and deliver the Rare Diseases Action Plan for Wales. The Network will consider how to increase grant income coming into Wales and how to increase the focus of building on existing collaborations across rare diseases research in Wales.

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Cross Cutting Areas

Functional Genomics

Wales has existing and growing strengths in functional genomics across Cardiff, Swansea, Aberystwyth and Bangor universities and this approach is already being applied across three main areas of genomics research activity in Wales: mental health, dementia and cancer, with single cell and spatial transcriptomics, and gene editing being the main areas of rapid expansion in Wales. Centres in Wales are associate members of the UK Human Functional Genomics Initiative. The centres using these advances include the UK Dementia Research Institute at Cardiff, the Neuroscience and Mental Health Innovation Institute, the Centre for Neuropsychiatric Genetics and Genomics, the Systems Immunity Research Institute, Cardiff University Division of Cancer and Genetics alongside Reproductive Biology and Gynaecological Oncology, and Immunobiology and Immunopathology in Swansea University. The combined grant income for research into functional genomics in Wales exceeds £66.6m. Other notable areas of activity include metabolomics using Welsh expertise in mass spectrometry (Aberystwyth and Swansea universities) and the Cardiff Systems Immunity Research Institute's work on IL-6 inhibition in rheumatoid arthritis that has led to the development of tocilizumab (anti-IL-6) and exploration of new blockers.

Pharmacogenomics

Pharmacogenomics expertise across Wales spans the translational research pipeline. Examples of excellence include: the Medicines Discovery Institute in Cardiff University employs around 40 people and has built a portfolio of projects to the value of £85 million to help address unmet need in mental health; Cardiff University's £9 million clinical pharmacogenomics research programme relating to the safety and efficacy of cancer treatments that has identified pharmacogenomic markers to improve treatment outcomes through personalised approaches to dosing and treatment; the £4 million funding in basic research into the biology of cancer at Bangor University led to the identification of pharmacogenomic markers and the development of several drug candidates via a spin-out company; and the £6 million programme exploring the health economic aspects of pharmacogenomics implementation and policy research at Bangor University has informed the adoption into practice of several single-gene tests relevant to the prescribing of treatments for diverse conditions (e.g. gout, HIV, epilepsy, colorectal cancer).

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In Swansea University, £4 million industrial-academic funding has catalysed the use of functional genomics to identify new disease specific targets and matched drug repurposing efforts in oncology and women's health.

Bangor University has also developed a health economic focus aspect to pharmacogenomics to guide clinical implementation, both within Wales and across the UK, which is well recognised within genomic UK networks such as the Centre of Excellence for Regulatory Science and Innovation Pharmacogenomics (CERSI-PGx). Clinical implementation of pharmacogenomic research is the main stay of pharmacogenomic service development and this is an area that is currently being explored at pace in Wales. It is recognised that for pharmacogenomic panel testing and its feasibility within clinical healthcare services, translational research is needed

to collect real-world data. Next steps are currently being scoped and defined within the Pharmacogenomic Delivery Plan for Wales developed by the National Pharmacogenomic Group (NPGG) where key actions and deliverables will be defined as part of the three-year plan aligned with this genomics research plan. Moving forward, genomics integration to trusted population databanks (such as SAIL databank) will broaden efforts, informing risk factors, predisposition, and clinical approaches for selective treatment regimens. Pharmacogenomics will be used increasingly to predict the likelihood that a particular medicine will be effective, and/or cause unintended harm through an adverse reaction. Through the existing research projects and a coordinated vision and approach defined by the pharmacogenomic delivery plan, Wales will be well placed to face this challenge.

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Capabilities In Wales

Infrastructure

Wales demonstrates capability in next generation sequencing and long-read sequencing, with core supporting technology for sample generation for single-cell and spatial transcriptomics, epigenomics and metabolomics. The existence of centralised capabilities at Canolfan Iechyd Genomig Cymru/ Wales Genomic Health Centre, All Wales Medical Genomics Service and Pathogen Genomics Unit, demonstrates Wales' commitment to investing in high-throughput genomic technologies.

All Wales Medical Genomics Service offers a variety of genetic testing services for the Welsh population including newborn bloodspot screening (from a heel prick test usually performed on babies five days after birth); the Wales Infants' and ChildreN's Genome Service (WINGS) (provides rapid whole genome sequencing service for seriously unwell children in neonatal or paediatric intensive care units); cancer genomic profiling for precision medicine; pharmacogenomics; and a wide range of rare and inherited disease services.

Pathogen Genomics Unit provides a number of genomic services including C. difficile, mycobacteria, HIV, SARS-CoV-2 and Influenza on an all-Wales basis. Public Health Wales plays a crucial role in leveraging genomics for disease prevention, outbreak response, treatment decisions and antimicrobial resistance monitoring. By collaborating with researchers through various strategic initiatives and partnerships such as their Public Health Wales Research and Evaluation Strategy 2023-2026, published in October 2023, they aim to enhance public health research and its practical applications.

The established infrastructure within Public Health Wales, All Wales Medical Genomics Service, Cardiff University and Swansea University provides a baseline for robust genomic data generation and processing. Genomics Partnership Wales has an objective to continue to enhance ways of working to ensure the best use of any laboratory equipment and technology. The co-location of genomic research and service partners at Canolfan Iechyd Genomig Cymru/Wales Genomic Health Centre provides significant opportunities for organisational collaboration. This integrated facility will deliver efficiencies to support future growth and advancement in genomics.

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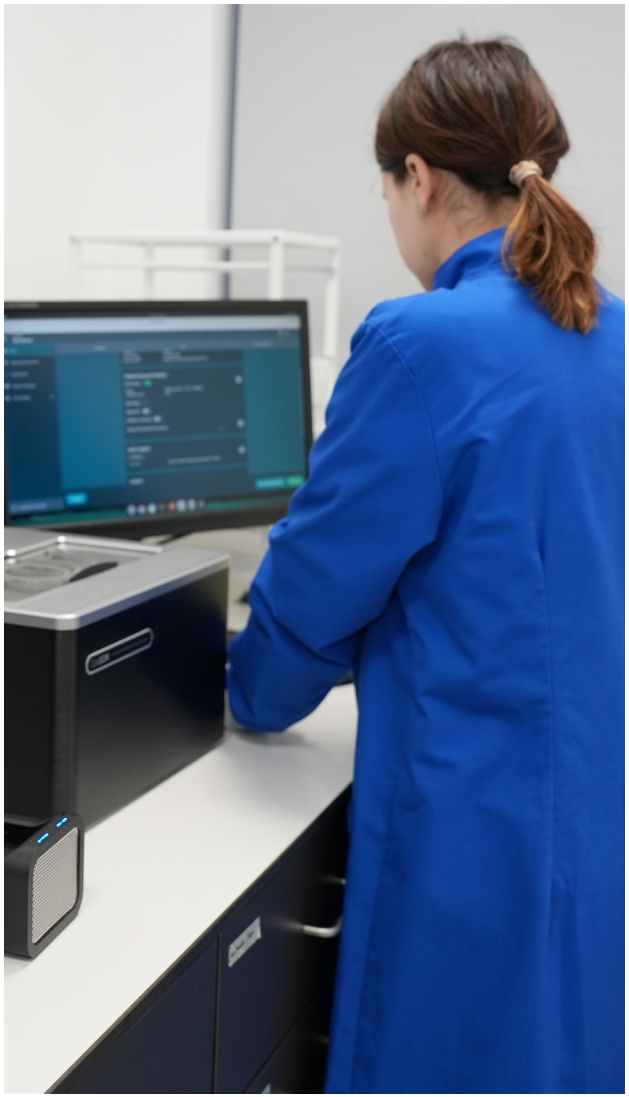

The BioResource Data Accelerator project is working closely with infrastructure groups such as the Wales Cancer Biobank, SAIL, Centre for Trials Research and Digital Health and Care Wales to leverage existing data, governance agreements and approaches to access consented and non-consented data, with a view to create and embed reproducible pipelines serving growing appetites for multimodal data and delivering on national research strategies.

Digital Health and Care Wales is advancing health and care research by integrating digital technologies and data insights. In 2023, it launched its first Research and Innovation Strategy, aiming to support impactful research, build partnerships, and foster innovation. Collaborating with NHS bodies, universities, and industry, Digital Health and Care Wales enables initiatives like the SAIL databank and are also working closely with Genomics Partnership Wales on the development of the digital delivery plan and roadmap for genomics.

Registries

The Welsh Cancer Intelligence and Surveillance Unit is the National Cancer Registry for Wales and plays a key role in supporting cancer research through its comprehensive population-level data, with records dating back to 1972. In addition to tracking cancer incidence, survival, and mortality, the Welsh Cancer Intelligence & Surveillance Unit enables the integration of somatic testing data for solid tumours and haematological malignancies, providing insight into the use and impact of precision medicine across Wales. Incorporating molecular testing data into cancer registration enhances population-level intelligence on tumour suitability for targeted therapies. Wales Cancer

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Intelligence and Surveillance Unit data is also shared with the SAIL Databank, creating opportunities for linked-data research and collaboration. Looking ahead, the collection of germline testing data offers further potential to explore the relationship between inherited genetic variants and tumour characteristics, helping identify high-risk variants and informing future screening and treatment strategies.

The Welsh Congenital Anomaly Register and Information Service was established in 1998 as a register of congenital anomalies in Wales. It has always included some rare diseases of genetic/hereditary origin and aims to provide reliable data on congenital anomalies which can be used to assess patterns of anomalies for 1998 to 2023. The Congenital Anomaly Register and Information Service includes conditions detected by the Newborn Bloodspot Screening Programme managed by All Wales Medical Genomics Service (as noted in the infrastructure section above). One of the functions of surveillance registries is to provide a data research platform. Quality of data is key therefore considerations around improving data flows (including cross border with England and other UK nations) and the use of GP data are of high priority. Research often emerges from clinical needs and therefore involvement of registries at all stages of research plan development will ensure asks are practical, possible and resourced.

Biobanking Resources and Pathology Services

Biobanks have provided both a vehicle and practical frameworks to enable genomic data sharing, which is an essential component in delivering this research plan. Partnering with Genomics Partnership Wales stakeholders, they also contribute to the development of technical solutions and computational infrastructure necessary for data sharing. Furthermore, the biobanks actively foster strong industrial partnerships, ensuring that maximum value is extracted from the sample and data collection efforts. Biobanking also plays a vital role in supporting the development of novel diagnostic tools and uncovering molecular mechanisms for rare diseases and cancer and are essential in diseases where samples are rare or difficult to obtain. Wales boasts a combination of well-established biobanks, such as the Wales Cancer Biobank and Cardiff University Biobank, alongside emerging initiatives like the All-Wales Genomic Databank. These biobanks specialise in the collection, processing, and archiving of genomic data and a wide range of patient-derived biological samples. This work relies on close

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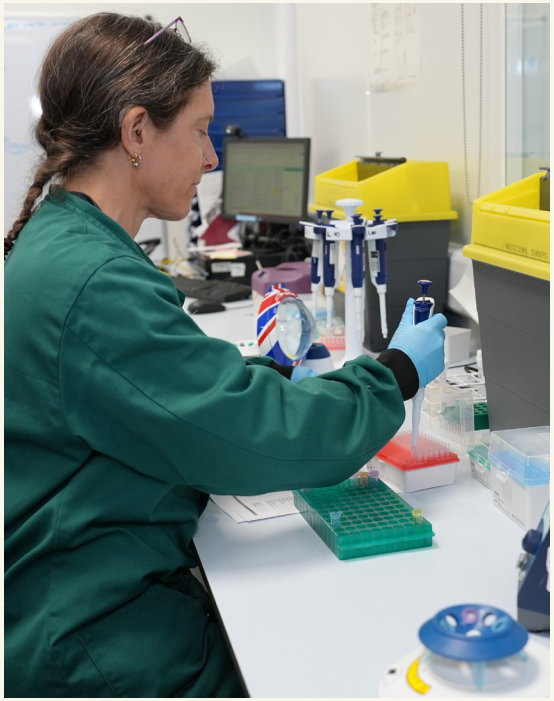
collaboration with pathology services and other clinical units to ensure both effective support and the timely collection of patient tissues (e.g. during surgery). Access to critical clinical information is integral to identifying disease mechanisms and improving diagnosis.

The Wales Cancer Biobank is a key infrastructure group committed to providing the operational framework to increase the capacity and capability for delivery of genomics-linked bio-samples to researchers, thus supporting the -omics data revolution in cancer research. This is being achieved through the Wales Cancer Biobank Digital project, which is establishing pipelines to enable access to diagnostic genomic data generated and held by the All-Wales Medical Genomics Service for patients who provide consent to Wales Cancer Biobank. This pipeline therefore complements and enriches the current data pipelines available to researchers complementing biosamples, image, sample and treatment/outcome data. Wales Cancer Biobank Digital, along with the other BioResource Data Accelerator project exemplars (e.g. All Wales Medical Genomics Service data into SAIL, and the use of the National Data Resource in conjunction with QuicDNA clinical trial data to undertake service evaluation) are directly delivering on national genomics strategies, plans, and commitments, including the Digital and data strategy for health and social care in Wales (2023) and the Genomics Delivery Plan for Wales (2022).

Broader 'Omics' and Translational Research Environment

There is a growing belief that combining genomics with proteomics and metabolomics



enhances disease prediction and understanding, especially in common and rare diseases. Protein signatures, often using a small number of markers, can outperform polygenic risk scores in predicting diseases, particularly cancers. While genetics identifies targets for gene

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therapy, proteomics reveals real-time protein changes, pointing to targets for existing drugs and repurposing opportunities and pharmacogenomics can incorporate multiple omics, especially proteomics, to better understand drug effects at varying doses.

Wales has leading Biological and Chemical Mass Spectrometry Facilities including Cardiff University Protein Technology Hub, Swansea University's NMSF and Lipidomics Laboratories, Natural Products BioHub and Aberystwyth University's High Resolution Metabolomics Laboratory. Swansea University also hosts part of the UK Rare Disease Research Platform and focuses on integrating mass spectrometry-based lipidomic science into rare disease research. This initiative aims to enable earlier diagnosis and improved clinical outcomes for patients with rare diseases.

One Wales Approach To Clinical Trials

The One Wales Support and Delivery Service provides coordinated, Wales-wide support for research sponsors and investigators, delivered through a national hub and local NHS R&D teams. It supports a wide range of studies, from early-phase trials to real-world observational research, by streamlining contracts, enabling adaptive delivery models, and providing access to expert staff and diverse patient populations. Delivered in alignment with UK-wide initiatives, Health and Care Research Wales is also developing Commercial Research Delivery Wales to enhance clinical trial capacity through the One Wales network. This includes providing targeted funding to scale up workforce and infrastructure for commercial research delivery via the Voluntary scheme for branded medicines Pricing, Access and Growth programme.

The Health and Care Research Wales funded Centre for Trials Research engages widely across the genomics research areas highlighted above and focus on the last critical step of clinical evaluation and future impact. For example, they support the QuicDNA and QuicDNA Max initiatives and work alongside teams across Wales in the mental health research area, including dementia and psychiatric disorders. They are a world leader in clinical trials in Huntington's disease and have pioneered work with the neurosciences team looking at the future of advanced therapies with direct implantation to the brain, with potential for genetic modification. The Centre for Trials Research also has significant expertise in use of routine data and data governance. Linking to pharmacogenomics, the growing role of genomic biomarker testing in both commercial and non-commercial clinical trials, particularly for targeted or

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advanced therapies has also been recognised as a priority. In addition, and in alignment with the cancer section above, highlighting how liquid biopsies and comprehensive genomic testing support trial eligibility, treatment selection, and monitoring provides valuable capabilities.

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Key Enablers

All Wales Medical Genomics Service

All Wales Medical Genomics Service has been performing genetic testing as a routine part of NHS patient care for over 30 years. Today, it delivers a wide range of genetic and genomic tests for thousands of patients, across the whole of Wales, every year and has collected a large archive of genetic data from the Welsh population. This data has enormous potential for health research. All Wales Medical Genomics Service plays a pivotal role in advancing genomic research across Wales by integrating clinical services, cutting-edge technology and collaborative partnerships. Being based at Canolfan Iechyd Genomic Cymru/ Wales Genomic Health Centre and having access to the newly refurbished laboratories and state of the art sequencing technologies there, All Wales Medical Genomics Service can foster collaboration among clinicians, researchers and industry partners, accelerating the translation of genomic discoveries into clinical applications.

Data For Research

Access to large genomic datasets is fundamentally important to genomic research. Realising the value of routinely generated data not only brings health benefits through the development of new treatments but also creates added economic benefits by encouraging the involvement of industrial partners. Wales' Genomics Delivery Plan committed to *"facilitate the appropriate and effective sharing of genomics data for research and innovation purposes"*.

SAIL Databank is a safe-haven for billions of person-based records, which enables researchers to answer important questions for the benefit of society by accessing a broad range of routine data collected over the last 30 years from an entire population. All Wales Medical Genomics Service and Pathogen Genomics Unit are the single service providers for standard of care genomic testing for human (cancer, rare disease and pharmacogenomics) and pathogens (HIV, TB etc.) respectively. Wales Gene Park, hosted by Cardiff University, supports genomic research and its application to healthcare. Wales Gene Park has a Data Integration team focused on improving flow of NHS genomic data to researchers. Facilitating an expansive data acquisition strategy to account for granular, regularly refreshed multi-modal data (including genomic, unstructured and imaging data) would provide the best way for Wales to sustain its competitive advantage in providing

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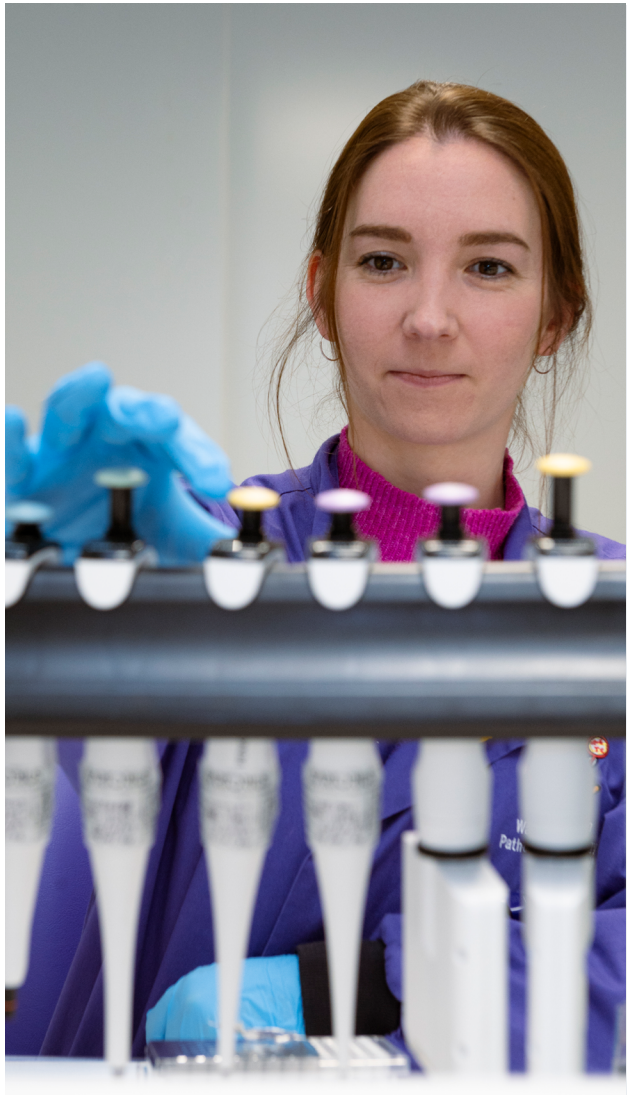
rapid, effective access to large scale datasets for health research. Building collaboration based on the strengths of SAIL databank, All Wales Medical Genomics Service, Wales Gene Park and Pathogen Genomics Unit to develop a shared vision for combining routine SAIL data with genomic data will enable Wales to maintain its leading position in health research data capabilities.

Consent For Research

The Lord O'Shaughnessy Review recommended that patients receiving genomic testing should be offered standard consent for engaging in research. This requires the development of an Information Governance policy and consent framework for genomic data that ensures data generated through direct care can be appropriately shared for research and innovation purposes. Work is also required to establish



a set of secure mechanisms so genomic data can be analysed alongside other clinically relevant data. This will include the creation of protocols and systems for the linkage and analysis of NHS and research genomic data with NHS data resources including SAIL databank. As detailed in the Genome UK Shared Commitments, collaborative working at a UK level around federated approaches to data access, best practice for data storage, and maintaining public trust and ethics for sharing personal data are also required. Through Genomics Partnership Wales Consent Group and appointed Consent Manager (a member of Wales Gene Park), and with ongoing involvement of Genomics Partnership Wales Patient and Public Sounding Board, the development of a research consent form and implementation framework is in progress.

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Patient Involvement

Patients and the public are acknowledged as partner in the genomics programme in Wales. Genomics Partnership Wales Patient and Public Sounding Board has been actively involved in many aspects of genomics in Wales through regular consultations. With diverse membership of lived experiences, the Sounding Board have co-produced published documents (including the Genomics Delivery Plan), provided input into many initiatives related to genomics, raised public awareness, and ensured patient-focused decision-making at the Board level within the programme. This ongoing partnership ensures that the patient voice is central to embedding genomics into Welsh healthcare and maintaining Wales's unique identity in this rapidly evolving field. In addition to supporting medical genetic and genomic research, Wales Gene Park has a strong track record in genomic education, engagement and involvement activities for both the public and healthcare professionals. It is well recognised that public and patient perspectives bring diverse insights and when effectively harnessed, lead to more nuanced and targeted research agendas.

Workforce

The workforce within the genomics research landscape in Wales shows distinct strengths (as highlighted above). There are also significant challenges due to labour market shortages across the UK driving demand for key skills; due to the competition for skills from the NHS and from industry; and due to data scientists not being able to move from a research environment into clinical bioinformatics as they are expected to have previous NHS experience. Therefore, there is a recognised need to work with the NHS, industry, and academia to develop new career frameworks and pathways to support those who wish to change or diversify careers.

Health Education Improvement Wales (HEIW) and Genomics Partnership Wales have recently developed the Strategic Workforce Plan for Genomics. This work acknowledges genomics as one of the fastest growing areas of medicine in the UK, recognises the current workforce challenges and details the actions that should be taken to ensure the workforce can deliver genomics in the future. One of the plan's key deliverables that is directly relevant to this research plan is: 'Development of clinical research in genomics through increasing the research and development workforce'. Genomics Partnership Wales are leading on the plan's actions in the strategic genomic workforce to: 'Establish the

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sustainable workforce infrastructure needed to facilitate the interface between the clinical and research environments, to realise both the research ambitions of genomics in Wales and the opportunity to attract strategic partnerships and industry investment' and 'Develop a pathfinder model to enable partnership arrangements with academic institutions and/or industry that allow staff within the specialist genomics workforce to span NHS service delivery and research/scholarship, to support the attraction and retention of skilled staff'.

Health and Care Research Wales produced a report in 2022: Making research careers work: a review of career pathways in health and social care in Wales which led to the development of the Health and Care Research Wales Faculty and a range of career awards that are open to individuals with an interest in pursuing a research career in genomics. The Healthcare Science Research and Innovation Strategy led by HEIW further outlines the importance of investing in research careers, training and development and the need for protected time for research for NHS staff. Close partnership between the implementation of the Genomics Research Plan and the Healthcare Science Research and Innovation Strategy will help to highlight and support the crucial and highly innovative Healthcare Science workforce.

Genomic research is driving the development of novel therapeutics, including gene and cell therapy, RNA-based therapies and gene editing technologies, broadly known as advanced therapies. Many advanced treatments will depend on genomic testing as part of the pathway of care. The number of new advanced therapies approved by regulators and evaluated by the National Institute for Care and Health Excellence is likely to accelerate over the next few years, increasing the number of genomic tests required. Working with Advanced Therapies Wales in this area as they develop their workforce as part of their Delivery Plan will be crucial.

Section Two: Taking Genomics Research in Wales Forward: What Are We Going To Do and How Will We Do It

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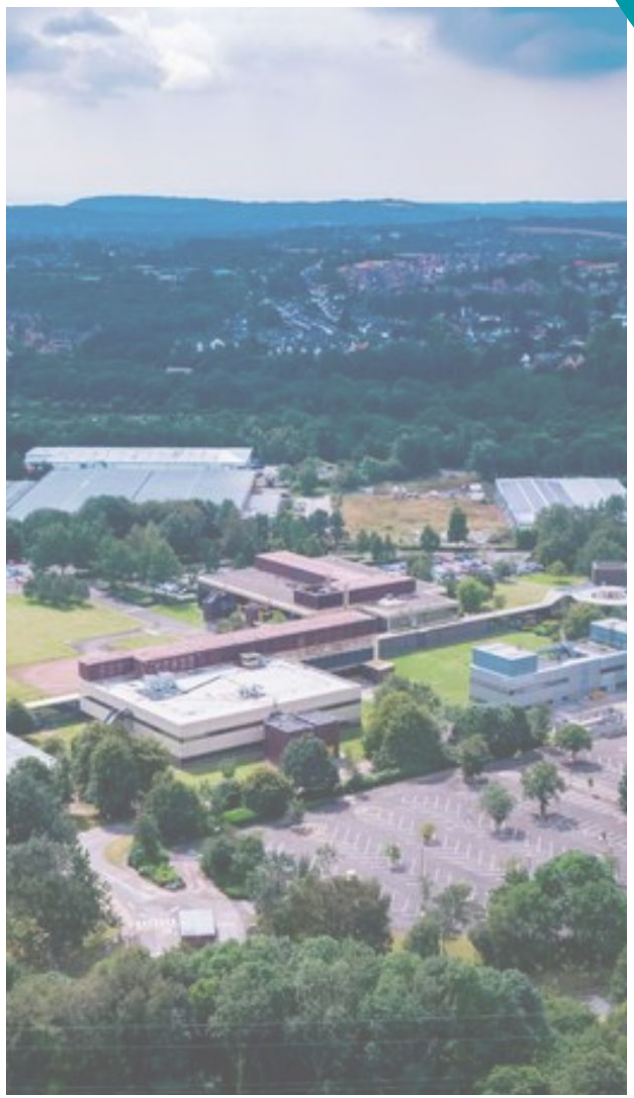
Wales is well-positioned to contribute to and benefit from national and international genomics research initiatives. This is evident in areas such as mental health, dementia, cancer, pathogen genomics, functional genomics, pharmacogenomics, and rare inherited diseases, as well as in key infrastructure elements including data, workforce, and patient/public involvement. However, while our strengths are acknowledged, it is also recognised that specific strategic objectives are needed to enhance our capabilities and support further development of the priority research themes outlined in the previous section of this document.

Given the rapidly evolving landscape, some of the actions required to fulfil these objectives may have clear tasks and timelines, while others will need further development and different timeframes. Similarly, some actions will require resources to be sought through applying for external grants, by exploring existing infrastructure funding and by securing industry partnerships. All of this will be considered at the implementation stage through a plan that will include consideration of funding pathways, governance structures, and measurable success metrics. Initially to kick start activities, the leadership role (Action 1.1) will be appointed (funded by Genomics Partnership Wales) to provide oversight and delivery as outlined below.



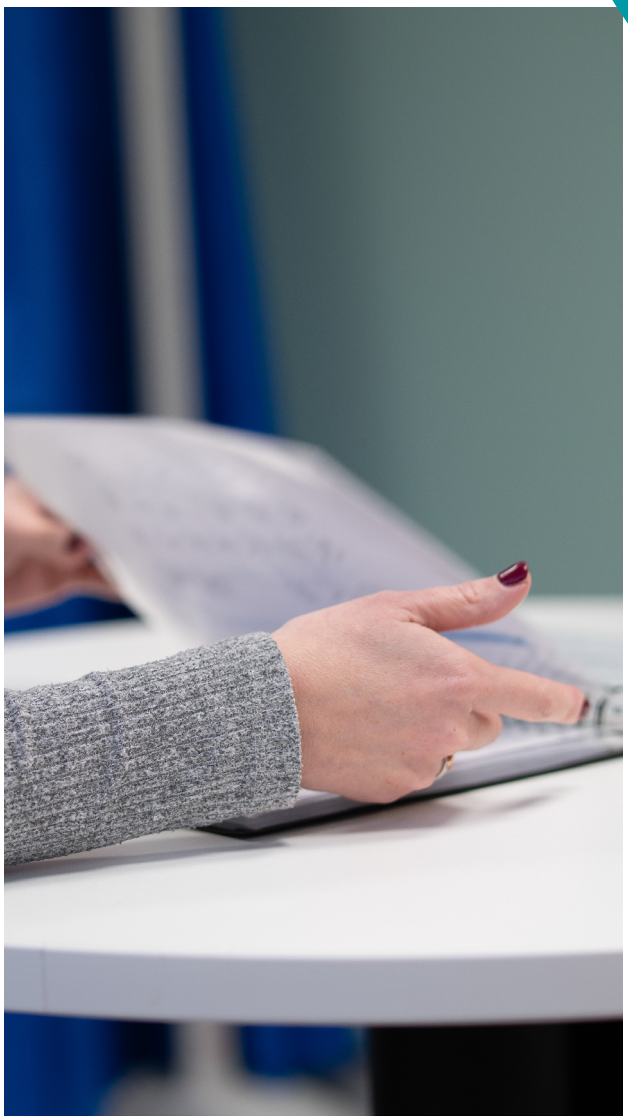
Section Two: Taking Genomics Research in Wales Forward: What Are We Going To Do and How Will We Do It

Immediate Strategic Objectives and Actions

Objective one: deliver a framework that brings together academia, NHS and industry to provide strategic leadership for genomics research in Wales.

- **Action 1.1** – Through close partnership between Welsh Government, Health and Care Research Wales, and Genomics Partnership Wales, we will establish a strategic leadership role to champion Welsh genomics research. **Welsh Government** and **Genomics Partnership Wales** [delivery: Autumn 2025]

- **Action 1.2** – We will refine the strategic advisory structure of the Genomics Partnership Wales' Research and Innovation Group, ensuring it has representatives from all partners, so that the Group can better support the strategic lead to develop genomics research that aligns with Welsh NHS priorities and needs, and strengthens and expands links with UK and global genomics initiatives. This will include ensuring a role and route for input from all the key units and activities identified through the research mapping **Genomics Partnership Wales** [delivery: December 2025]



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Objective two: build on this framework to grow the genomics research community in Wales and establish it as a hub for collaborative genomics research.

- **Action 2.1** - We will investigate and test ways to support the development of Wales-led multidisciplinary collaborations to access funding, attract talent and drive advancements in emerging research areas such as the development and use of Artificial Intelligence. **Health and Care Research Wales** [delivery: ongoing]

- **Action 2.2** - We will seek to retain and concentrate the talent we have in genomics research by providing high quality work environments through the identification of funding sources and supporting applications to help continue successful projects such as CLIMB and QuicDNA. In addition, we will help create new investments in less well funded areas such as rare diseases and collaborative and translational research. **Health and Care Research Wales** [delivery: ongoing]

- **Action 2.3** - We will raise awareness and visibility through a Welsh national genomics event to complement the UK's Festival of Genomics or Genomics England's research symposia and encourage Welsh researchers to put themselves forward for the UK events. **Genomics Partnership Wales** [delivery: annual or biennial event]

- **Action 2.4** - We will develop and implement a clear plan for fostering public-private partnerships, offering incentives for industry-led research, and positioning Wales as a leader in real-world genomic data analysis. We will explore ways to enhance collaboration with industry to drive innovation and translate research into practical applications via initiatives such as industry-academia partnerships and shared research projects. **Health and Care Research Wales** [delivery: ongoing]

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- **Action 2.5** - We will develop and implement a national public engagement strategy that encompasses not only the Genomics Partnership Wales Sounding Board but also the broader public, ensuring a more inclusive and extensive public conversation by safeguarding independence and targeting representation from wider groups such as ethnically diverse, LGBTQIA+, other protected characteristics groups as well as taking into account socio-economic status including people from rural settings, older people and people with a disability. **Genomics Partnership Wales** [delivery: December 2025]

- **Action 2.6** - We will actively explore and exploit funding opportunities to accelerate genomic driven trials with commercial partners and the VPAG investment programme to improve patient access to commercial clinical trials. Similarly, we will consider the needs of registries at all stages of relevant research plans to ensure appropriate funding and workforce are considered. **Health and Care Research Wales** [delivery: throughout 2025 and 2026]

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Long-Term Strategic Objectives and Actions

Objective three: establish the key building blocks for the sharing and use of genomic data (human and pathogen) in Wales.

- **Action 3.1** - Building on existing work and expertise in data sharing, both within Wales and across the four nations, we will develop a vision for the safe and pro-innovation use of data in research. This will address Wales-wide agreements on information governance, regulation, and consent; the technical and operational infrastructure needed to support data sharing; and the required resources across clinical and research teams. For rare diseases specifically, any Welsh National Genomics Research Library we will ensure alignment with the English equivalent. In addition, we will use learning from these activities to inform work on the sharing of routinely collected tissue samples for research. **Genomics Partnership Wales.** [delivery: 2026]

- **Action 3.2** - Building on the work that is already ongoing in Wales in organisations such as Wales Cancer Bank and Digital Health and Care Wales, we will develop a single national NHS genomic research consent mechanism that is clear, efficient, and adaptable. The model will be scalable and easily integrated into activities across the NHS. **Genomics Partnership Wales** [delivery: by the end of 2026]

- **Action 3.3** - A defined role within UK consortia could enhance Wales' access to cutting-edge genomic datasets, large-scale clinical trials, and national funding opportunities. We will encourage the involvement of Welsh stakeholders, as collaborators or leaders, in large scale UK-wide projects such as Our Future Health. **Health and Care Research Wales** [delivery: ongoing]

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- **Action 3.4** – We will work towards broadening genomic data collection to include diverse populations, ensuring equitable access to genomic testing and research participation. **Genomics Partnership Wales** [delivery: ongoing]

Objective four: ensure that Wales is equipped with the required computational infrastructure and analytical expertise to exploit the data that is generated in Wales for genomics research.

- **Action 4.1** – We will coordinate with Digital Health and Care Wales through the genomic digital delivery plan activities, as well as more broadly across life sciences, to understand computational infrastructure needs for genomics research in Wales to build a cohesive ecosystem. **Genomics Partnership Wales** [delivery: align with digital strategy]

- **Action 4.2** – We will work with NHS partners to understand parallels between their computational infrastructure, the research infrastructure and the broader community to help inform funding requests and decisions and assist with future planning. **Genomics Partnership Wales** [delivery: ongoing]

- **Action 4.3** – We will coordinate with the universities and health boards to gain an understanding of the local specialties and expertise to develop an idea of the skills shortages that may exist in Wales (e.g. in bioinformatics and bioengineering in particular) and understand which organisations are best placed to help fill those shortages. We will support the specific research-related actions listed in the Strategic Workforce Plan for Genomics, the Healthcare Science Research and Innovation Strategy, and Advanced Therapies Wales Delivery Plan to ensure that the workforce is in place with the required skills to support genomics research in Wales. **Health Education and Improvement Wales** [delivery: ongoing]

Appendix 1

Name	Role
Kieran Walshe (Chair)	Director Health and Care Research Wales and Chief Research Advisor to CMO, Welsh Government
Carys Thomas (Chair)	Head of Research and Development Policy, Welsh Government
Delyth Morgan	Head of Life Sciences Research and Industry Partnerships, Welsh Government
Alex Newberry	Head of Research Governance, Public Involvement & Digital Research, Welsh Government
Andrew Fry	Clinical Senior Lecturer in Medical Genetics at Cardiff University and Honorary Consultant Clinical Geneticist in the All Wales Medical Genomics Service
Clive Morgan	Managing Director, All Wales Medical Genomics Service
Dyfrig Hughes	Professor of Pharmacoeconomics, Bangor University
James Walters	Director, Centre for Neuropsychiatric Genetics and Genomics, Cardiff University
Jemima Foy	Senior Health Science Policy Manager, Welsh Government
Laura Bunting	Research Infrastructure and Experimental Medicine Lead, Welsh Government
Leon Wong	Deputy Director Health Science, Welsh Government
Michaela John	Head of Programme, Genomics Partnership Wales
Nick O'Sullivan	Programme Manager, Genomics Partnership Wales
Nigel Williams	Professor, Division of Psychological Medicine and Clinical Neurosciences, Cardiff University
Sian Morgan	Director of Laboratory, All Wales Medical Genomics Service
Simon Thompson	Co-Director SAIL, Swansea University
Steve Conlan	Professor of Molecular and Cell Biology, Swansea University and Chair of the GPW Research and Innovation Working Group.
Tom Connor	Professor Cardiff University and Public Health Wales Pathogen Genomics Unit.

In addition to the Task and Finish group and wider consultation work, Genomics Partnership Wales Patient and Public Sounding Board played a key role in shaping and drafting the plan.