Llywodraeth Cymru
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GIG CYMRU NHS WALES

Partneriaeth Genomeg Cymru
Genomics Partnership Wales

Genomics Delivery Plan for Wales

2022 – 2025

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Contents

Foreword

Genomics is amongst the most exciting arenas of 21st century healthcare and will give us previously unimaginable opportunities to better understand illness, improve patient outcomes and transform lives.

Personalised medicine, testing and other genetic adaptations and innovations using new methods, tools and technologies will shape the future of modern medicine.

The Genomics Delivery Plan for Wales 2022-25 details how Genomics Partnership Wales will continue to work in partnership 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.

We will develop skills of our existing workforce so that a modern generation of healthcare professionals may emerge with the capabilities to seize the opportunity for the people of Wales.

A person-centred, joined-up approach will enable us to create and take opportunities right across healthcare, and we look forward to supporting the plan to deliver upon our ambitions to improve health, wellbeing and prosperity.

The development of a new state of the art genomics facility at Cardiff Edge Life Sciences Park for 2023 will physically bring together partners who deliver services and research, as well as the supporting infrastructure, to create an environment which promotes the sharing of knowledge and expertise.

We would like to express our thanks to Genomics Partnership Wales' Patient and Public Sounding Board for their continual dedication and support in shaping the way genomics is being delivered now and into the future.

Eluned Morgan MS
Minister for Health and Social Services

Suzanne Rankin
Senior Responsible Officer, Genomics Partnership Wales

Putting the patient's voice at the heart of genomics policy

We are members of the public with lived experience of genetic conditions or genetic testing as part of our healthcare, and are given the chance to inform decision making and influence the direction of genomics in Wales; this pioneering approach has proven to be highly effective.

Established in 2019, the Sounding Board has around twenty members from diverse backgrounds and with a broad range of experiences.

Working closely with Genomics Partnership Wales colleagues from several disciplines and organisations, we have co-produced some key elements of the genomics programme; this Delivery Plan, designs for the new building at Cardiff Edge, the Genomics Showcase conference, and the Wales Rare Diseases Action Plan. We have been involved in service development and research consent approaches. We are actively involved in raising awareness of Genomics and two of our members sit on both the Genomics Partnership Wales Governance and Programme Boards, ensuring transparency and involvement in decision-making at the highest level.

Genomic medicine is expanding apace with new research bringing almost weekly breakthroughs, it is expected to become a pivotal strand of healthcare. In order to exploit these opportunities for faster and more accurate diagnosis, it needs to be embedded into many core speciality areas including cancer, paediatrics, neurology and mental health.

This will require all health boards and trusts in Wales to enable genomics education within their workforce as key part of their professional training. This is critical to ensure that the benefits reach the population across communities in Wales. It is important to work collaboratively in genomics across the UK, but it is also vital that we do not lose our own identity here in Wales, to ensure that our patients, population and economy benefit.

The good work achieved so far by the Sounding Board must continue with the patient voice retained as an important element in the development of genomics in Wales in the long term.

Genomics Partnership Wales Patient and Public Sounding Board

The Sounding board will continue to be of paramount importance in improving what Wales can offer those people and their families/carers with Rare Diseases by sharing their wide range of experiences, insights and ideas – Dr Jamie Duckers, Clinical Lead for Rare Disease in Wales.

Genomics in Wales: Impact for our patients and population

Gene tests that helps Welsh patients avoid chemotherapy

Cancer is a disease of the genome and the Cymru Service for Genomic Oncology DIagnoses service means we're now able to expand our genomic testing for patients with cancer. Up until now we've only been able to detect genetic changes in around 30 genes, but the service means we're now able to detect changes in around 500 genes. We can also test many more types of tumours, so our service at the moment involves testing lung cancers, colorectal cancers, melanomas and a couple more tumours – Rhian White, Head cancer genomics clinical scientist.

Partneriaeth Genomeg Cymru
Genomics Partnership Wales

More details:
https://medicalgenomicswales.co.uk/index.php/health-professional-information/cysgodi
https://www.bbc.co.uk/news/uk-wales-58654240

Genetic testing: Babies with rare illnesses given hope

We've had an answer to around four out of 10 cases. That's really important considering when I started my career about 30 years ago we were lucky to get an answer in one in 10 of the tests we were doing then. That's because the technology has completely changed in recent years. WINGS makes all the difference to the families in terms of diagnosis and treatment. We know without diagnosis these people are coming back to the health service again and again for more tests – Sian Morgan, Head of Laboratories.

WINGS
Wales Infants' & ChildreN's Genome Service

More details:
https://medicalgenomicswales.co.uk/index.php/health-professional-information/wings
https://medicalgenomicswales.co.uk/index.php/homepage/professional-news/162-awmgs-and-wings-staff-make-debut-on-primetime-bbc-wales-today-news
https://www.bbc.co.uk/news/uk-wales-61062322

Mental Health – Genetics Service to help patients and families

This service represents the realisation of many years of research in Cardiff and across the world to find out more about the contribution of genetics to the risk of developing mental illnesses. For some people, genetics plays an important part in the development of their mental health problems, and we hope this new service can contribute to improving their mental and physical health. We are one of a few teams in the world offering this service and it's exciting to be setting out on this journey with patients, their families and colleagues from genetics and mental health services in the Welsh NHS – James Walters, Consultant Psychiatrist and Professor in Psychiatry at Cardiff University.

Pharmacogenetics

Wales became the first in the UK to routinely provide all cancer patients being treated with certain types of chemotherapy DPYD screening to identify their risk of severe side effects and help prevent this occurring.

As doctors and health care professionals we are tasked with improving the well being of our patients and avoiding doing harm. Over the last 20 years I have seen and cared for many patients who have benefitted from this treatment but also looked after those who have suffered the more severe side effects, with more drastic consequences. This test now allows us to assess the risk of these side effects and in selected patients to change the treatment to reduce the likelihood of this occurring. DPYD screening allows us to improve the quality of care of cancer patients across Wales and save lives – Richard Adams, Professor and Consultant in Bowel Cancer at Velindre and Cardiff University.

More details:

https://www.bowelcanceruk.org.uk/news-and-blogs/news/patients-in-wales-to-receive-routine-life-saving-testing-ahead-of-chemotherapy-treatment/

https://medicalgenomicswales.co.uk/index.php/about/pharmacogenetics

SECTION ONE

Setting the scene: why are we doing this and where are we now?

Overview

Genomics is revolutionising the way we think about healthcare. It is providing us with a far more detailed understanding of what causes illness and infectious disease and is underpinning the development of new interventions that would have been unobtainable a decade ago.

We are at an important juncture in the history of genomic healthcare. Rapidly decreasing sequencing costs, combined with increased computing power mean we are able to understand the human genetic code like never before. Using genomic data, we are able to enable new ways of delivering services and creating new opportunities for public health action. We are well-placed to harness advances in our understanding of genomics to respond quickly to evolving threats, including COVID-19.

Building on the intentions of **A Healthier Wales: Our Plan for Health and Social Care**, in September 2020, Wales signed as a partner the UK Government's strategy **Genome UK: The Future of Healthcare**. This ten-year strategy sets out the ambition for the UK to have a world-leading genomic healthcare system for the benefit of patients.

In March 2022, we committed to working collectively with our UK partners to progress our objectives over the next three years; to deliver better health outcomes, work towards the UK being the best location globally to conduct genomics research and to grow new genomics healthcare companies.

Genomic advancements involve both devolved and reserved functions of government; for example, health is devolved whilst other areas such as ethics and Medicines & Healthcare products Regulatory Agency are not. **Genome UK: Shared Commitments for UK-wide implementation 2022 to 2025** details how the four nations will work together for the next three years, and this delivery plan expands on how we will deliver genomics for Wales during this period.

This plan forms a key part of our ambitions to transform diagnostics services and redesign pathways, and will link to the Diagnostics Recovery and Transformation Strategy for Wales (2022-25) which is currently being developed.

Journey so far/ Our Genomics Ecosystem

In Wales, we enter this new phase of our genomics story from a position of strength in partnership.
In 2016, we issued a **Statement of Intent for genomics and precision medicine in Wales**, outlining our plan to use new genomics technologies to improve the health and prosperity of the people of Wales.

2017, we launched the Genomics for Precision Medicine Strategy for Wales, providing more detail and practical steps aligned to the statement of intent.

In 2018, Genomics Partnership Wales was formed to deliver the **Genomics for Precision Medicine Strategy**. Our key partners are represented below:

OUR KEY PARTNERS

We supported our strategy with significant investment and over the past few years we have seen this investment bear fruit, with advancements in genomics in several key areas within Wales. Genomics Partnership Wales' strengths include the cross-organisational approach, close alignment to policy and a demonstrable ability to work with agility and adapt at pace.

The Partnership has enabled new technologies to be adopted and deliver real benefits for patients with the introduction of new services; the Wales Infants' and ChildreN's Genome Service has received 52 referrals in the first two years, with a third of these critically ill patients receiving a diagnosis, allowing them to receive the correct care and treatments; first in the UK to routinely provide screening for all cancer patients being treated with certain types of chemotherapy to identify their risk of severe side effects, with around 4,000 patients tested within the first two years, nearly 7% identified as at risk, and given alternative treatments; the Cymru Service for Genomic Oncology DIagnoses, (CYSGODI) delivers the increasing demand for cancer biomarkers for precision medicine molecular pathology services for cancer patients, and in the first nine months, 1,967 patients received testing with 542 having actionable genomic alterations leading to personalised cancer treatments and better outcomes.

The Partnership has established Genomics Cafes across Wales for members of the public to find out about new advances in genomic medicine in Wales. Prior to the pandemic, these were local events with an average of 15 attendees; once in-person events were not possible, they were delivered online and attendance numbers increased significantly. They continue virtually every six weeks with around 50-100 attendees.

The inaugural Genomics Showcase event in 2021 was a huge success with 1,200 individuals registering for the virtual conference, both members of the public and professionals, and future annual events are planned. Since 2018, over 30 Genomics Roadshows have taken place at hospitals across Wales reaching around 900 health professionals, educating, informing and empowering future service development.

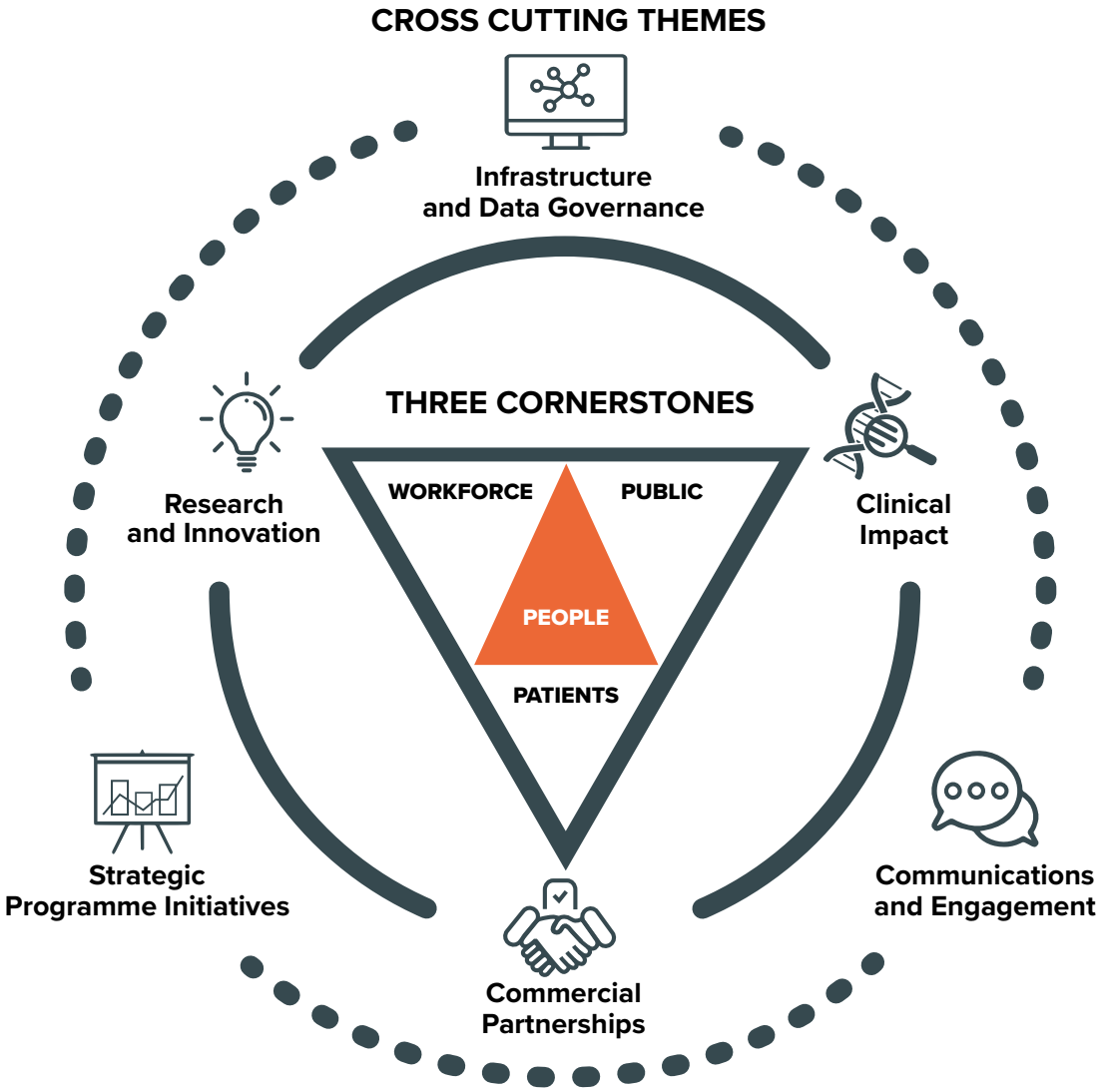
The pandemic has focused a spotlight on pathogen genomics and demonstrated the significant benefits of building on strong foundations we have in Wales. Throughout the pandemic the Welsh team sequenced and shared in excess of 200,000 viral genomes placing Wales in the top ten globally and had a direct impact on patients and our wider society. These genomes have underpinned surveillance efforts relating to SARS-CoV-2 variants and mutations of concern in Wales and the UK; the genomics data has been used in real time to support over 100 outbreak investigations in workplaces, hospitals and prisons, in order for control measures to be put in place; data has been routinely generated and shared with health boards and government, to support pandemic planning and decision making. The routine and transparent sharing of the SARS-CoV-2 genomic data internationally has enabled the global research community to advance overall knowledge of SARS-CoV-2, and other countries have looked at Wales to guide and inform their own response locally. The data shared has been used in over 50 international publications. Software to make genomic data 'usable', written by bioinformaticians in Wales, has been used worldwide to analyse millions of SARS-CoV-2 genomes.

This is not happenstance, but a product of investing in people and our services such that they can develop and thrive.

Using the intentions of the Well-being of Future Generations (Wales) Act of 'working better with people and communities and each other, look to prevent problems and take a more joined-up approach', all of the activity outlined above has been supported by our Patient and Public Sounding Board. Our Genomics Ecosystem in Wales is founded upon placing people at the centre of our work. It commits to being inclusive of all genomics strengths in Wales whilst enhancing our outward-facing approach to develop exciting collaborations.

Our ambition via this Plan is to put us in a position to use genomics to transform healthcare delivery and public health action in the future, focusing on building capability and enabling opportunity around genomics across all of the relevant sectors, for the benefit of our patients and the population of Wales.

OUR GENOMICS ECOSYSTEM IN WALES



Our vision/ Guiding principles/ Delivery themes

Our vision –

'Working together to harness the potential of genomics to improve the health, wellbeing and prosperity of the people of Wales'.

To support our vision, our priorities are as follows:

- We will continue and progress our **programme of communication and engagement** with the public. We will ensure that meaningful patient and public involvement shapes what we can do with genomics in Wales.
- We will create a '**Genomic Centre for Wales**', making Wales a great place for NHS, academia and industry to work in genomics. It will be connected to a network of regional delivery 'nodes', ensuring that patients across Wales benefit fully from genomics as part of their healthcare and that opportunities to work in our genomic services are distributed across Wales.
- We will **sequence up to 3,000 whole genomes annually within the next 3 years**, allowing us to offer more extensive genomic testing and support to patients with a suspected rare disease and for specific cancer types thus shortening the diagnostic journeys, enabling effective treatments and improving outcomes.
- We will commit to offer up to **5,000 extensive genomic testing profiles** to patients with newly diagnosed cancer annually ensuring improved diagnosis in order to improve cancer outcomes.
- As part of future pandemic planning, we will continue to **strengthen our genomic surveillance of pathogens** to support public health response and policy making; while **continuing to build world-leading diagnostic services for pathogen genomics**.
- We will continue to **develop and invest in our healthcare workforce** by training and supporting them to acquire the relevant knowledge and skills so that they can deliver the benefits of genomics to their patients through improved clinical pathways and standards of care.
- We will commit to developing and **supporting research and innovation** across areas of strength in our higher education institutions, health boards and other centres of excellence.
- We will develop a **sustainable and secure storage solution** for genomic data and samples, with appropriate data governance, ensuring genomic data can support the health and care of the people in Wales alongside a thriving research environment.
- We will work in partnership with **Advanced Therapies Wales** to develop the necessary requirements for delivering gene therapy trials and approved gene therapy products.
- We will continue to **work with our UK partners** to progress and achieve our main objective over the next three years; to deliver better health outcomes across the UK.

- The UK strategy highlights commitments set out across the three pillars of Genome UK – diagnosis and personalised medicine, prevention and early detection, and research; and the five cross-cutting themes – ethics and maintaining trust, engagement and dialogue with patients and the public, data, workforce development and industry.

Recognising our own approach within Wales, our delivery plan is split into the following four key theme areas:

- **Delivery Theme 1:**
  A Focus on People.
- **Delivery Theme 2:**
  Clinical Services (Genome UK Pillar 1 &2).
- **Delivery Theme 3:**
  Research and Innovation (Genome UK Pillar 3).
- **Delivery Theme 4:**
  Enablers (Genome UK Cross-cutting themes).

Delivering what really matters to our patients and the public is central to our ambition and vision for genomics in Wales. Our Genomics Partnership Wales Patient and Public Sounding Board has developed guiding principles that will serve as the foundation for our future delivery; this includes our commitment to communicating clearly with patients and involving them in prioritising deliverables to ensure maximum benefit; to provide high quality inclusive and accessible services with appropriate levels of support; timely diagnoses, appropriate treatment and best care for patients and their families.

We will act with integrity, transparency and ensure that the Welsh population will benefit from our actions both now and into the future.

SECTION TWO

What are we going to do and why are we going to do it?

Delivery Theme 1: A Focus on People

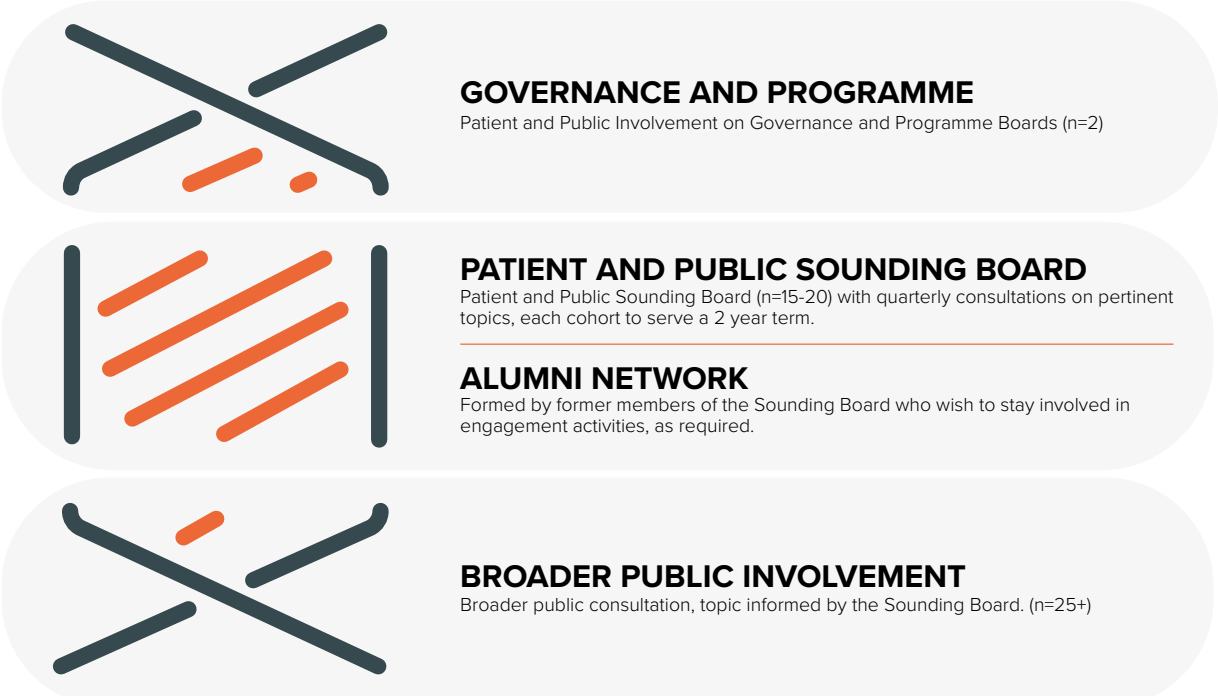
Patient and Public Involvement/ Co-production

In Wales we are committed to:

'work in an open and transparent manner with patients and the public, using their collective experiences to shape and add value to the work of the Genomics Partnership and future genomics services in Wales'.

Strong foundations have been laid for patient and public involvement and co-production, with a three-tiered approach established to ensure a broad range of opportunities for patients and the public to work with us to strengthen the quality of genomics output:

OUR THREE TIERED APPROACH TO CO-PRODUCTION



We will continue to build on our expertise and experience to evolve this key aspect of our work with ongoing dedicated budget, the continued support from all Genomics Partnership Wales members and alignment to the Wales Gene Park engagement programme. We recognise that increased engagement with communities is key to strengthening the patient 'voice' in the development of genomics; we will continue to regularly recruit patients and members of the public to work with us from diverse groups, recognising the strength of a membership which provides a good reflection of the overall Welsh population, including an increased focus on involvement of young people and underrepresented communities within Wales.

Our ambition is to further develop our approach to involvement and to embed it as a core function of genomics services and research activity in Wales.

To support our approach to patient and public involvement we will...

Deliverables:

- Use this Plan to develop public information to promote genomics ambitions.
- Grow the 'alumni' (former members of the Sounding Board) community of patients and the public with involvement in specific areas of the genomics work including evaluation of progress.
- Create a suite of resources and establish our Sounding Board and alumni members as community ambassadors to raise the profile of genomics and how individuals will benefit.
- Create a strategy which demonstrates how to transition to a model where patient and public involvement becomes a permanent function of, and 'in-house' to, service delivery.
- Develop an evaluation strategy for the genomics programme using the co-production framework to demonstrate the impact and benefit of our activities.
- Create links between the involvement work in genomics and other patient groups and initiatives around healthcare involvement both within Wales and across the UK.
- Create an appropriate forum to involve more young people in the genomics programme.

Workforce (The specialist, non-specialist and academic workforce)

Attracting, developing and retaining a highly-skilled, motivated and dedicated genomics workforce is a high priority for ensuring our vision for genomics is achieved within Wales.

This includes ensuring that clear career routes exist for scientific and clinical roles within the human and pathogen specialties.

A specific objective in the 2017 Strategy was to increase further Scientist Training Programmes and Higher Specialist Scientist Training posts in genetic counselling, bioinformatics, genomic science, microbiology and molecular pathology, and specialist training posts for Clinical Geneticists, as required; much of this was achieved.

We recognise that no advancement in technology or new equipment in isolation can benefit patients, and investment in the workforce is vital. This is particularly critical for workforce groups where there are national shortages. Engagement and support from senior leaders across the NHS will be crucial in enabling us to empower and up-skill the non-genomics healthcare workforce. This is vital to the integration of genomics into the patient pathways where clinical care is improved.

The previous Strategy identified the requirement to determine training needs of the wider healthcare and academic workforce in genomics and to provide appropriate training opportunities for NHS Wales staff.

Work in this area, through specialty-focused surveys, found that the vast majority of the workforce is poorly informed about genomics, and many do not recognise its importance and/or relevance. To address this, MSc Genomic Medicine courses were established, and funded, part-time places provided for NHS staff; subsequently a more modular approach was taken to reach a larger number of individuals. An online 'basic genomics' resource is also being developed and will soon be available through Health Education and Improvement Wales's e-learning platform. Many continuing professional development events and opportunities have been provided, the majority of which coincided with the launch of new services.

Bioinformatics is essential for the translation of raw genomic data into healthcare and public health benefit, and commercial opportunities. Our vision is for Wales to nurture and grow a world-beating bioinformatics workforce and associated knowledge assets, supported by the appropriate digital technologies, to deliver on the service, research and innovation objectives.

More broadly, this includes the need to attract and retain high quality data scientists, and software and system engineers in the genomics area by developing an appealing career structure which competes with other sectors. It is important to acknowledge that these are new areas of activity for the NHS, which require specific planning and consideration.

We also need to ensure that we retain specialist genomics expertise in Wales and ensure transition of critical members of temporary staff on to permanent contracts to nurture genomic excellence in Wales.

To support our approach to workforce we will need...

Deliverables:

- Aligned to a wider diagnostics workforce plan, a comprehensive and strategic genomics workforce plan developed by Health Education and Improvement Wales and Gene Park Wales Workforce and Training group. This plan will include:
  - the support of our specialist genomic workforce to meet increased demand for genomics, which aligns to Health Education and Improvement Wales' Healthcare Science and other workforce plans; increasing staff numbers, including implementing a plan for the development of the data/digital workforce, and considering other ways to increase capacity to meet patient demand such as extended working hours for the clinical and diagnostic service;
  - details and a set of tools to upskill the wider healthcare workforce in genomics that may include:
    i. Providing new training packages;
    ii. Identifying a dedicated genomics education and training officer in every hospital/postgraduate centre;
    iii. Setting up a network of dedicated genomics quality leads across Wales;
    iv. Ensuring there are national genomics leads for core specialties and alignment to workforce plans across relevant areas including oncology, pharmacy, cardiology, psychiatry, paediatrics, neurology, ophthalmology;
    v. Supporting staff to be both confident and competent with regard to taking consent for genomic tests and interpreting the results as well as discussing the implications of the results with patients;
  - the development of clinical research in genomics by increasing the Research and Development workforce, including the creation of new clinical and non-clinical lecturer and/or senior lecturer posts, high-level degree studentships and post-doctoral research officer positions;
  - links to UK-wide workforce and education initiatives as part of the UK Shared Commitments with collaboration through the Genomics Training Academy.

Communication and Engagement

Communication and engagement activity will serve as a cross-cutting theme within our genomics ecosystem and will underpin all aspects of delivery.

We will continue to take an all-Wales approach to genomics communication activities, ensuring that the work of the Partnership is promoted to all audiences across Wales and internationally, and that these audiences are engaged via the most appropriate channels.

This will be supported by the Wales Gene Park who have an established education and engagement programme around genomics for health professionals, researchers, schools and colleges, people affected by rare and genetic conditions and the wider public; opportunities to establish further networks will be explored.

To support our approach to communication and engagement we will...

Deliverables:

- Develop the Genomics Partnership Wales Communications Strategy further to support and endorse the communication and engagement activities of the individual organisations as well as promoting the collective work of the Partnership model.

- Raise awareness of genetics and genomics through broader public outreach and engagement across all communities, focusing on under-represented audiences and those affected by health inequalities, informed by good patient and public involvement and providing information in accessible formats.
- Raise awareness of genetics and genomics amongst the existing and future healthcare workforce.
- Develop a new Research Involvement Initiative to promote patient participation in research and clinical trials, and supporting genomics researchers with their education, engagement and involvement activities.
- Continue to support Genetic Alliance UK in several initiatives including the development of a Welsh Action Plan to implement the UK Rare Diseases Framework, the development of the adult and paediatric Syndromes Without a Name (SWAN) clinics in Wales and contributing genomic expertise to the various relevant Welsh Cross-Party Groups including Rare, Genetic and Undiagnosed Conditions, and Cancer.

Delivery Theme 2: Clinical Services (Genome UK Pillar 1 & 2)

Human – All Wales Medical Genomics Service

Human genomic medicine for rare disease, cancer and pharmacogenetics is a rapidly evolving field with the capability to transform healthcare outcomes through improving prevention, diagnosis and treatment of disease. Rapid advances in treatments using gene therapy will require an integrated approach, where genomics will underpin treatments. Personalised medicine will be an integral and vital aspect of future mainstream healthcare in the NHS in Wales, incorporating and embracing the latest in genomic advances, and bringing the greatest benefit to our patients.

Genomic laboratory services are delivered for the Welsh population by the All Wales Medical Genomics Service. In April 2020, the All Wales Medical Genomics Service introduced rapid whole genome sequencing in new-born and paediatric intensive care units allowing infants to receive a diagnosis faster and reduce the ‘diagnostic odyssey’, to improve access to innovative treatments and specialist care. Taking forward the Wales rare diseases action plan and priorities, we will continue to build upon recent investment in genomic diagnostic technologies and expand whole genome sequencing to improve diagnostic rates.

Wales was the first UK nation to introduce Non-Invasive Prenatal Testing as a contingent test in the NHS antenatal screening pathway. We will continue to invest in prevention and early detection genomics-based programmes in order to improve outcomes for babies and their families through safe and effective early diagnosis.

We will establish a collaborative public health system in Wales that uses genomics to strengthen screening, diagnostic and care pathways in those at high risk. We will also enable and encourage lifelong good mental health and wellbeing by anticipating, predicting and acting to improve outcomes in those at increased genomic risk of poor mental health and associated poor physical health.

COVID-19 has negatively impacted cancer care in Wales and there is an urgent need to expand routine use of diagnostic genomic testing in order to improve cancer outcomes. In 2021 the All Wales Medical Genomics Service launched the large 500 multi-gene panel to deliver precision medicine services for cancer in Wales.
The next ambition is not only to further maximise identification of molecularly matched therapies at the point of cancer diagnosis for better patient cancer outcomes in Wales, including development of whole cancer genomes, but also rapidly implement the increased utilisation of liquid biopsy within the cancer pathways to inform treatment earlier, and also to detect relapse earlier and more effectively, for screening and, earlier diagnosis and surveillance.

Adoption of pharmacogenomics within NHS Wales has already become a reality with the delivery of the DPYD gene screening in 2020, enabling the most effective provision of tailored therapies to individual patients. Expanding pharmacogenomics will not only lead to improved outcomes for the patient but also seeks to reduce adverse drug reactions, which have a considerable impact on the NHS.

Our ambition is to support and drive the further development of pharmacogenomics, by developing services that provide timely information relevant to prescribers. Specifically, the availability of pharmacogenomics information that is important for optimising many commonly-prescribed medicines will lead to improved health outcomes for patients.

Ensuring that the right dose of the right drug is selected for the right patient at the right time will reduce the burden of adverse drug reactions, and improve the effectiveness of treatments. The pharmacogenomics service will be delivered in an equitable and cost-effective manner.

To support our approach to human genomics we will...

Deliverables:

Cancer: Realising the promise of precision medicine

- Offer more comprehensive genomic profiling to patients with newly diagnosed cancer so that by 2023, up to 5,000 patients a year can access these tests in Wales.
- Establish a task and finish group to ensure that there is a coordinated national approach with defined academic and clinical evidence-led input for the adoption of liquid biopsy for the screening, diagnosis and surveillance of cancer.
- Continue to implement liquid biopsy within the NHS Wales national cancer optimal pathways to identify patients with clinically actionable gene targets (personalised treatment).
- Support and deliver genomic-enabled early clinical cancer trials, for example where it will be essential in the delivery of gene therapies.
- Through the All Wales Genomics Oncology Group support the by further developing and strengthening established links between the All Wales Medical Genomics Service, Clinical Oncology, Cellular Pathology, Haematology, Pharmacy, Wales Cancer Network, Welsh Health Specialised Services Committee, All Wales Medicines Strategy Group and All Wales Therapeutics and Toxicology Centre.

Rare and Inherited Diseases: Realising the promise of increased diagnosis for Rare Disease

- Roll-out of whole genome and exome sequencing to patients with a suspected rare disease to accelerate diagnosis. Our ambition is to sequence up to 3,000 genomes annually within the next 3 years with a final target of delivering 5,000 genomes within the next 5 years.
- Develop a repatriation strategy for Specialist Rare Disease Services (from NHS England back to NHS Wales). Our ambition is to re-patriate up to 2,500 tests.
- Evaluate the activity of genes (the function of most genes is not yet known) through the development of transcriptomic/metabolomics technologies, which will speed up definitive diagnosis and reduce the rare disease 'diagnostic odyssey'.
- Develop long read sequencing capacity to further increase the number of patients identified with a Rare Disease and reduce the 'diagnostic odyssey'.

Developing an NHS pharmacogenomics service for Wales

- Develop a pharmacogenomics plan which will outline the processes for the effective implementation of the service, and which recognises the importance of multi-professional education and training, clinical pathway development, information technology and laboratory practice.
- Establish the 'National Pharmacogenomics Group' to ensure that there is a multidisciplinary, coordinated national approach with defined clinical and academic input to the development and introduction of pharmacogenetic services in Wales.
- Establish, within the All Wales Medical Genomics Service, a cost-effective pharmacogene panel service to cover multiple pharmacogenetic targets to inform the present and future prescribing needs.
- Develop appropriate decision-support tools for doctors and pharmacists within electronic health records.

Prevention and early detection: vital ambitions for the healthcare system in Wales

- Expand non-invasive prenatal testing to other reproductive pathways to improve patient outcomes and optimise resource utilisation.
- In collaboration with the Wales Screening Committee, Public Health Wales and Newborn Bloodspot Screening Wales, develop an evidence-based plan to identify where genomic technologies can be used to improve accuracy and timeliness of newborn screening services, enabling early detection and quicker diagnosis of conditions where early intervention provides better outcomes for patients.
- In collaboration with the public health and screening system in Wales, explore the potential uses of genomics to strengthen the current biochemical screening, diagnostic and care pathways.

Mental Health

- Develop and expand the All Wales Psychiatric Genomics Service, including establishing pathways and protocols within mental health services for referral of patients, and providing genetic counselling for these individuals and their families.

Advanced Therapeutic Medicinal Products

- Continue to strengthen the partnership between the Genomics Partnership Wales and Advanced Therapies Wales to ensure that there is the genomic testing capacity required to support and advance the ambitions of the **Advanced Therapies Statement of Intent** and developments in gene therapy.

Pathogen – Pathogen Genomics Unit, Public Health Wales

Pathogen genomics is transforming how we deliver healthcare in Wales, from patient to population. Pathogen genomics offers data that can be directly integrated into public health analysis and decision-making, while simultaneously enabling precision healthcare. It therefore enables us to offer a new way of working - Precision Public Health, integrating, in real-time, the diagnosis and characterisation of pathogens that infect individuals, with efforts to prevent disease and control outbreaks on a population level.

Wales already occupies a world-leading position for the delivery of genomics services focused on SARS-CoV-2, C. difficile, HIV and Influenza. We also work with UK Health Security Agency to deliver world-leading genomics services for Tuberculosis and Non-tuberculosis Mycobacteria.

Pathogen genomics has almost limitless potential, with thousands of potential pathogens that could be sequenced. There has been a significant increase in the sequencing capacity, the size and capability of the pathogen genomics workforce, and information/analysis systems in Wales during the pandemic. This critical mass provides the required experience of delivering transformational genomics services to underpin an ambitious programme to mainline pathogen genomics into the NHS in Wales over the coming years.

The Pathogen genomics team in Wales have built a modular sequencing and analysis platform, which lowers the barrier to implementing and operating new services. As the need for SARS-CoV-2 sequencing reduces, this modular approach provides the opportunity for the introduction of future pathogen services.

The ambition is to build on the established, accredited clinical pathogen genomics services; to continue to work with stakeholders across Wales to improve patient and well-being objectives for population health by focusing on three key areas:

- Provide surveillance.
- Use pathogen genomics to support the development of new services to improve healthcare.
- Public health crisis response.

These focus areas align to a number of Public Health Wales strategic priorities including:

- Protecting the public from infection and environmental threats to health.
- Supporting the development of a sustainable health and care system focused on prevention and early intervention.
- Building and mobilising knowledge and skills to improve health and well-being.

To provide a framework for developing and embedding pathogen genomics as a business-as-usual capability within our healthcare system, we will...

Deliverables:

Work to mainstream Pathogen Genomics through two key deliverables, which will provide a framework for developing and embedding pathogen genomics as a business-as-usual capability within our healthcare system.

These deliverables are:

- Embed genomics as part of the core vision for Public Health Wales, with the creation of a cross-organisational programme for Public Health Genomics within Public Health Wales.
- Development of a Pathogen Genomics delivery plan for Wales, co-produced will stakeholders; it will provide a roadmap for the Public Health Genomics programme and provide an agreed framework for the evolution of pathogen genomics services in Wales. Those elements which will be included as a minimum are detailed in Annex 1.

Delivery Theme 3: Research and Innovation (Genome UK Pillar 3)

Research and innovation in the field of genomics and precision medicine is advancing at scale and great pace, worldwide. Genomics research and innovation in Wales has established strengths in cancer, dementia, mental health, rare disease and pathogen genomics. More recent and ongoing developments include liquid biopsy, long-read sequencing, pharmacogenomics, functional genomics (single cell and spatial transcriptomics, and epigenomics) and new-born screening; areas that are receiving substantial interest and investment, and where Wales has the potential to emerge as a UK leader, including delivering clinical trials.

Research and innovation are being undertaken in Welsh Universities and their Medical Schools in Cardiff (which hosts Wales Gene Park), Swansea, and Bangor, together with NHS Wales Health Boards (including All Wales Medical Genomics Service hosted by Cardiff and Vale University Health Board). Current centres will be enhanced by the Welsh Government's investments in a new genomics centre at Cardiff Edge Life Sciences Park, and two new major pathology centres in South West and South East Wales that seek to integrate innovations in genomics.

Genomics research and innovation is integral to the Cancer Research Strategy for Wales and aligns with priorities in the NHS Wales Precision Medicine Advanced Therapies Wales initiative, and the Wales rare diseases action plan. Genomics research and innovation will also underpin future clinical trials capabilities and deliver opportunities in Wales.

Health and Care Research Wales has made major investments in genomics through the Wales Gene Park, that supports researchers through facilitating access to genomic sequencing technologies and providing analytical expertise. Wales Gene Park also works to inform and engage patients, public and professionals about development in genomic research. Short term Wales Gene Park priorities include: (i) increasing bioinformatic and data science support for researchers, and (ii) facilitating researcher access to clinical genomic data and samples routinely generated by All Wales Medical Genomics Service.

Intrinsically linked to research and innovation is access to a wealth of other human, pathogen and functional genomic data, generated in Wales' university research centres, and to UK genomic data repositories such as Genomics England Research Environment, UK Biobank, CLIMB and recently SAIL. Genomic researchers in Wales will also contribute to and utilise data from future UK genomics research initiatives including Our Future Health and the Genomics England Newborn Genomes Programme.

An initial Health and Care Research Wales-led genomics research and innovation appraisal has identified the genomics of mental health, dementia, cancer, and pathogens, functional genomics, pharmacogenomics, and rare and inherited disease genomics as key areas on which to focus the future genomics research and innovation strategy for Wales. Opportunities to further enhance genomics research and innovation will require investment in both clinical and non-clinical academic workforce, working in those research centres delivering the genomics research and innovation strategy.

To support and further develop our research ambitions, we will...

Deliverables:

- Develop a genomics research strategy for Wales, including:
  - — Undertaking a review of the genomics research environment in Wales (including defining research remit/scope across translational pipeline, engagement and consultation with stakeholders and the public).
  - — Reviewing genomics infrastructure research needs in Wales.
  - — Establishing an external advisory group to support and underpin the review and strategy development work.
- Develop an independent, objective assessment for future financial genomics research investments, including:
  - — Assessing current Welsh Government research investments/strategic plans.
  - — Reviewing the Welsh Government genomics research investments (e.g. Wales Gene Park, All Wales Medical Genomics Service research services).
- Establish processes to enable 'find, recruit and follow-up' systems to support genomics-enabled clinical trials and research studies.
- Through the Wales Gene Park:
  - — Increase the number of bioinformaticians employed in Wales;
  - — Facilitate researcher access to routine NHS genomic data through a consent system and development of a trusted research environment;
  - — Continue education and engagement activities.

SECTION THREE

How we support our delivery now and into the future?

Delivery Theme 4: Enablers (Genome UK Cross-cutting themes)

In order for the ambitions within this Genomics Delivery Plan to be realised, including genomics activities from research through to the provision of clinical services, there are a number of key enablers fundamental to providing the necessary foundation.

Infrastructure

Premises

We are providing £15 million to co-locate All Wales Medical Genomics Service, Pathogen Genomics Unit and Wales Gene Park at a purpose-built facility at Cardiff Edge Business Park. This co-location will integrate research and clinical service delivery in the field of genomics. It will provide modern, scalable laboratories, patient clinics and working areas that encourage collaboration to drive innovation and keep Wales at the leading edge of genomic service delivery and research.

The creation of a national genomics centre on the Cardiff Edge site will be the cornerstone of a potential Precision Medicine Centre of Excellence, allowing further collaboration between NHS Wales, academia and Industry.

The national genomics centre will be strengthened by a robust network of 'nodes' situated initially in South West and North Wales, linking into the existing national life science infrastructure and considerably strengthening the precision medicine ecosystem in Wales. This provides exciting opportunities and will deliver a significant contribution to our aim of supporting and developing research excellence, which positively impacts the health, wellbeing and prosperity of the people in Wales.

In order to support this, we will...

Deliverables:

- Co-locate Wales Gene Park, Public Health Wales' Pathogen Genomics Unit/Public Health Genomics Programme and All Wales Medical Genomics Service in a single, fit-for-purpose building at Cardiff Edge.

Equipment

Significant investment has been provided to increase sequencing capacity across the genomics community in Wales; in the last 12 months additional instrumentation has been implemented to increase clinical service provision across both the human and pathogen services.

To ensure we have the appropriate equipment to support our ambitions in Wales we will...

Deliverables:

- Develop a national governance framework and implementation infrastructure for the rapid adoption of novel and emergent genomic technologies.
- Develop a forward plan to identify additional sequencing infrastructure, robotic platforms and sample preparation required in Wales.
- Continue to enhance ways of working to ensure the best use of any laboratory equipment. This will include identifying opportunities for organisational collaboration with shared instrumentation, automation, data analysis and storage capabilities.
- Maximise opportunities for synergies between genomics and other disciplines such as pathology services and medical oncology.

Digital and Data

A key aim for this plan is to develop a sustainable and secure solution for the storage, management and access of genomic data, supported by appropriate data governance and digital technology. Work has already taken place to support the expansion of genomics over the course of the first strategy; to build upon this and accelerate development in this critical area, a digital architect has been appointed to support the evaluation and delivery of future technical requirements. This will include the need to underpin and modernise clinical services, including making genomics results part of the single patient record; build synergy across the partnership to share best practice, maximise return on investment from sequencer estate, compute and storage, and develop a shared support model.

DIGITAL AND DATA SUPPORT MODEL

To ensure genomic data can support the health and care of the people in Wales and a thriving research environment we will...

Deliverables:

- Implement the required Local Area Network and Wide Area Network solutions at Cardiff Edge to support the three partner organisations' genomics activity.
- Develop a strategy for the sustainable and secure storage, management and access of genomic data that will maximise its use for patient and public benefit in Wales. This will include the development of a data archive storage plan where we will assess the benefits/options of shared storage requirements at Cardiff Edge.
- Develop an Information Governance policy for genomic data that ensures the use and sharing of data for direct care purposes, and facilitates the appropriate and effective sharing of data for research and innovation purposes with on-going monitoring and evaluation to meet latest legal requirements.

- Establish a set of secure mechanisms so genomic data can be analysed alongside other clinically relevant data. This will include the establishment of protocols and systems for the linkage and analysis of NHS and research genomics data with NHS data resources including SAIL.
- Develop and implement a digital roadmap that will support our data strategy, including networking infrastructure to allow unhindered flow of genomic data between NHS, academia, and where appropriate, industry.

Commercial Partnerships

A fundamental ambition of the Genomics for Precision Medicine Strategy was not only to drive better health outcomes but also to become an international destination for genomics and precision health research; to attract global commercial partners and to create jobs and wealth for the population of Wales.

The vision for Wales to become internationally recognised as a hotspot for precision medicine genomics is dependent on the continuing development of an ecosystem consisting of businesses working with cutting-edge technology and collaborating with world-class research, small and mid-size enterprises, and industry partners.

To encourage relocation of Industry and increase inward investment, Wales must be seen as an approachable nation, open for business and ready for collaboration. We need to ensure that we maintain the devolved independence, agility and unique benefits that we provide for collaboration:

- Close to policy makers.
- Population size and stability.
- Genomics Partnership Wales' structure for Wales-wide partnership working.
- Wales as a great place to live and work.

To support our approach to commercial partnerships we will...

Deliverables:

- Develop an international strategic marketing plan aligned with the wider life sciences strategy which:
  - creates an online presence for genomics in Wales which clearly describes what the national offer is and highlights all of the achievements to recognise the expertise of the NHS genomics workforce.
  - builds upon a comprehensive precision medicine global sectoral analysis to determine what the best genomic health commercial partnership offer is for Wales;
  - encourages collaboration for mutual benefit with commercial partners to invest in Wales;
  - allows NHS organisations in Wales to be less commercially risk averse.
- Align with the Welsh Government Health and Care Innovation programme, including:
  - enabling commercial development capability and capacity;
  - integrating with the Welsh Innovation Ecosystem;
  - offering partnering opportunities at scale and pace.
- Support the development of a centre of excellence for genomics, to:
  - develop an NHS/academic master-plan for Cardiff Edge site at pace as part of the national precision medicine infrastructure;
  - support life science developments;
  - encourage a pipeline of ideas created in Wales as a global 'shop window'.

Cellular Pathology

To meet the ambitions within the plan the All Wales Medical Genomics Services will require cellular pathology capacity provision to match genomic cancer testing targets. In order to achieve this, we will...

Deliverables:

- Work with the National Pathology Programme and the National Diagnostics Programme to develop a Cellular Pathology Plan to support future genomic testing, and ensure that the implementation of the **Pathology Statement of Intent** incorporates the requirements for genomics.

Funding and Commissioning

Over the past five years, Welsh Government has provided significant funding and invested in genomics in Wales.

Genomics is currently a specialist service. To ensure that the population of Wales has fair and equitable access to the full range of specialised services, in 2010 the Welsh Health Specialised Services Committee was established. They are responsible for the joint planning of specialised services on behalf of the Health Boards in Wales to reduce any possible duplication and ensure consistency.

For All Wales Medical Genomics Service, the current genomics service specification and commissioning policy was updated 2022, which sets out the criteria and clinical circumstances required for patients to have access to a genomics service. Having an appropriate commissioning policy is key to ensure:

- an equitable, high-quality and sustainable genomics service for the population of Wales;
- that genomics services are evidence-based with demonstrable benefits for patient care and management;
- a service is delivered efficiently and provides timely information to patients, families and clinical teams;
- a service will support timely and evidence-based access to new therapies;
- a service provides value for money to commissioners and to the public.
- patient and public involvement / co-production in genomics.

To support our approach to commissioning we will...

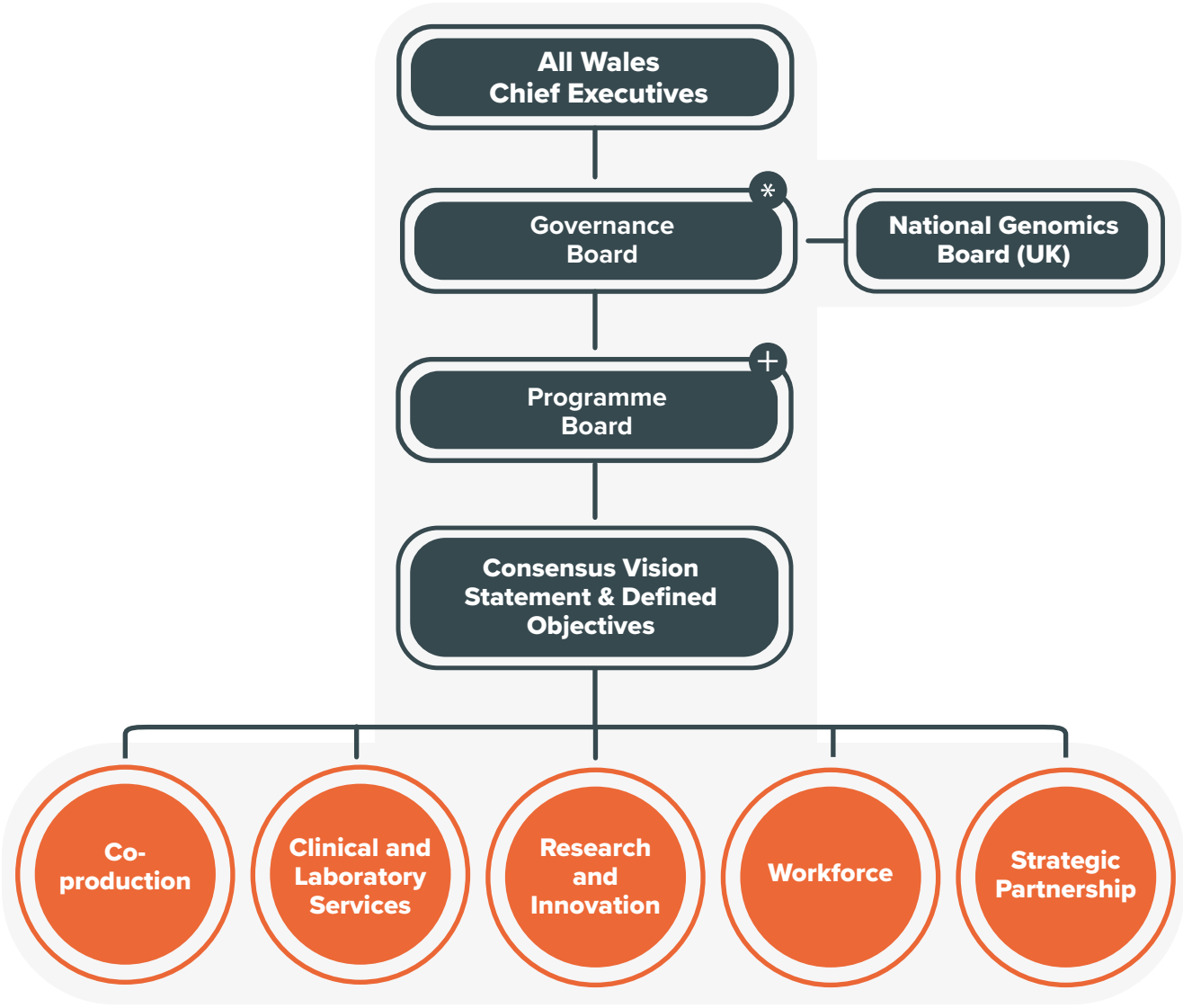
Deliverables:

- Revise and update the current genomics commissioning policy in Wales, including;
  - confirming the scope of commissioned genetic tests in the context of a phased increase in availability;
  - ensuring alignment with NHS England's test directories to maintain equity for the population of Wales with the rest of the UK;
  - developing a process to commission specific evidence-based tests that may be outside of the test directories.
- Develop a demand and capacity plan for the expansion of services required to deliver pathogen and public health genomics; pharmacogenomics; new-born screening programmes; rare diseases; cancer services including symptomatic and screening; and medical genetics.
- Develop an enabling resource framework to support the delivery plan using the principles of Value Based Health Care.

SECTION FOUR

How will we know what we have done, how successful we have been and ensure that we are ready for the future?

GOVERNANCE



* As described in our programme for transforming and modernising planned care, diagnostics programmes will be brought together under a National Diagnostics Board to support a holistic approach towards earlier diagnosis and timely interventions.

\+ Responsible for understanding and managing the impact of change, owning the resolution of risks, representing local strategy and resolving dependencies with other pieces of work.

Deliverable:

- To review the governance structure in line with the new Delivery Plan.

Horizon scanning

Advances in our understanding of genomics are happening at significant pace and are enabling changes in how health services can be developed and delivered. It is therefore vitally important, that we keep abreast of these advancements in genomics so we can ensure we are adequately prepared to take advantage of the opportunities to improve the services we provide.

Horizon scanning in genomics is already included within the Welsh Health Specialised Services Committee horizon scanning process. It includes drawing on national sources, such as National Institute for Health and Care Excellence appraisal intentions, horizon scanning undertaken by other agencies, advice from the All Wales Medical Genomics Service through its horizon scanning and Welsh Health Specialised Services Committee own horizon scanning work.

For public health genomics, Pathogen Genomics Unit also undertakes horizon scanning activities.

To support our approach to horizon scanning we will...

Deliverable:

- Develop an approach to genomics horizon scanning including the establishment of an implementation group with relevant stakeholders.

Evaluation

Genomics Partnership Wales identified from the outset the importance of a robust evaluation strategy to underpin the delivery of genomics in Wales, to ensure that progress was quantifiable and that a culture of continuous improvement was embedded within our ways of working.

Pre-pandemic, initial scoping work was undertaken with a combined NHS-academic research organisation (Cedar) to develop an independently-verified evaluation plan, and to undertake a uniform approach across all Precision Medicine Programmes.

To ensure we have a robust approach we will...

Deliverable:

- Develop an evaluation and monitoring strategy for the programme using the involvement framework, and Sounding Board and alumni (former Sounding Board members) members, to demonstrate the impact of the programme and to provide a mechanism to review progress against aims.

ANNEX 1

Pathogen Genomics Delivery Plan for Wales – The Pathogen Genomics Unit, Public Health Wales

Development of a Pathogen Genomics delivery plan for Wales, which will bring together views from across stakeholders in Wales to provide a masterplan for the development and improvement of pathogen genomics services in Wales.

The delivery plan will provide a roadmap for the Public Health Genomics programme and provide an agreed framework for the evolution of pathogen genomics services in Wales. This delivery plan will include the following elements:

- Roadmaps to improve and enhance the delivery of our current pathogen genomics services focused on SARS-CoV-2, C. difficile, HIV and Influenza, including improving turnaround times and increasing the actionable information from existing services.
- Specific pandemic planning preparations, including the development of a new accredited metagenomics service to enable the characterisation of unknown pathogens in patient samples, and the provision of analysis systems for pandemic preparedness.
- Engagement with stakeholders across Wales to identify and plan out opportunities for transformational services that genomics enables, for example in sexual health, which could be developed and delivered in Wales.
- Building on existing UK-wide reference laboratory services delivered in Wales to add genomics services for Cryptosporidum, Anaerobes and Mycology which could be offered out beyond Wales.
- Establish a virtual genomic surveillance team as part of the Public Health Genomics programme to plan and deliver genomics surveillance services, and to provide a core capability for the integration of genomics into future pandemic and epidemic responses.
- Integrate genomics into sentinel surveillance activities for respiratory pathogens, with a roadmap for adding new species for genomic surveillance.
- Continue to work with colleagues from across the UK to deliver world-leading genomics services for Tuberculosis and Non-tuberculosis Mycobacteria.
- Work with colleagues from across the UK to identify, evaluate and, where appropriate, use pathogen genomics services provided by the NHS and other public health agencies.
- Work with colleagues from across the UK to design and implement digital systems to enable the use of services elsewhere in the UK, and to provide a route for our services to be available to other parts of the UK.
- Work directly with bioinformatics teams across the UK public health agencies to build shared analysis pipelines to provide a true ‘federated’ bioinformatics capability, managing, sharing and co-creating analysis pipelines in an integrated and collaborative way with other UK public health agencies.

- Develop a shared vision for shared digital infrastructure and collaborative working with human genomics, identifying ways to build infrastructure and overcome joint challenges such as those relating to data sharing across the UK.
- Develop a plan for the delivery of one health genomics services, working with human genomics to link host and pathogen genomics, and working with external stakeholders across the UK to understand pathogens in the environment and how this impacts human health.
- Work with international stakeholders to support the development of agreed standards and best practice for pathogen genomics.