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Wales Gene Park

Annual Report 2022-2023



Ymchwil Iechyd
a Gofal Cymru
Health and Care
Research Wales



Afiennir gan
Lywodraeth Cymru
Funded by
Welsh Government

2022 - 2023
Annual report



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WELCOME

Welcome to the Wales Gene Park annual report which summarises Wales Gene Park activities during the period 1st April 2022- 31st March 2023.

This year marks the 20th anniversary of the founding of Wales Gene Park. Over the past two decades Wales Gene Park has supported research, innovation and healthcare in Wales through advances in genomic sequencing, developing novel laboratory methods and facilitating hundreds of cutting-edge research projects.

This annual report captures the value and impact of the work undertaken by the Wales Gene Park. We are working hard to deliver key aspects of the Genomics Delivery Plan for Wales 2022-25. To do this, Wales Gene Park has a broad portfolio of activities that include an extensive programme of engagement with patients and the public, education of medical professionals, providing researchers access to advanced genomic technologies, and bespoke support with bioinformatic analysis.

Wales Gene Park is an integral part of Genomics Partnership Wales, and we are moving imminently to co-locate with the NHS All Wales Medical Genomics Service and the Pathogen Genomics Unit of Public Health Wales at the new Genomic Centre in Coryton. We look forward to the opportunities and challenges ahead!



Angela Burgess, Education & Engagement Lead/Co-Director, Wales Gene Park

FOREWORD

This annual report captures the value and impact of the broad portfolio of work undertaken by the Wales Gene Park team.

A major role of Wales Gene Park is to make genomic technologies available to biomedical researchers in Wales. In the last 12 months the Wales Gene Park has generated sequencing data for 68 genomic projects, provided bioinformatics support for 20 projects and supported external research grants bringing in over £3.42M to Wales. Wales Gene Park provides bespoke support for critical areas of research - cancer, rare disease, immunology, and others – including many projects that could not be accommodated by larger commercial organisations.

Another key goal of Wales Gene Park is to engage with the public to promote understanding of the opportunities and challenges raised by genomics. We are working hard with the public and patient representatives to develop secure and appropriate strategies for sharing routinely collected genomic data to empower health research.

Genomics is a fast-moving field and Wales Gene Park continues to evolve and adapt. This year we said goodbye to Dr Karen Reed. Karen had been the Wales Gene Park Operations Manager since 2017. It was a privilege to work with Karen and we wish her well in her future career.



Dr Andrew Fry, Director, Wales Gene Park

A WORD FROM OUR PATIENT AND PUBLIC REPRESENTATIVES

It really is reassuring that Wales Gene Park puts the public at the heart of everything it does and that it strives to ensure diverse public and patient involvement and engagement. The wide range of public engagement events, including talks at community organisations, genomics cafes and public lectures demonstrate that Wales Gene Park is committed to ensuring that public involvement and engagement is accessible and meaningful. I am delighted to see all the good work that has already happened and am even more excited about the work that is planned for the future.

Mr Nathan Davies, Wales Gene Park Patient & Public Representative

I am very proud to be part of the Wales Gene Park, as a patient it is good to be a part of a forward-looking organisation and having input that effects the “*end user*”, the patient. The huge amount, of work that WGP achieves is very impressive.

Mr Alan Thomas, Wales Gene Park Patient & Public Representative

INTRODUCTION & STRATEGIC OBJECTIVES

Introduction

Wales Gene Park is funded by the Welsh Government through Health and Care Research Wales. We support, promote, and perform medical genetic and genomic research of the highest quality and work to ensure its translation for the benefit of patients and public in Wales and beyond.

Mission

- To promote and facilitate Welsh medical genetic and genomic research and its application to improve health and wealth in Wales.
- To engage the public and health professionals to improve understanding of the opportunities and challenges arising through genetics and genomics.

Aims

Wales Gene Park works to support the implementation of the Welsh Government's Genomics for Precision Medicine Strategy by:

- Promoting and facilitating high quality genetic and genomic health research in Wales.
- Ensuring the informed involvement of patients, public and professionals in the development of genomic medicine in Wales.

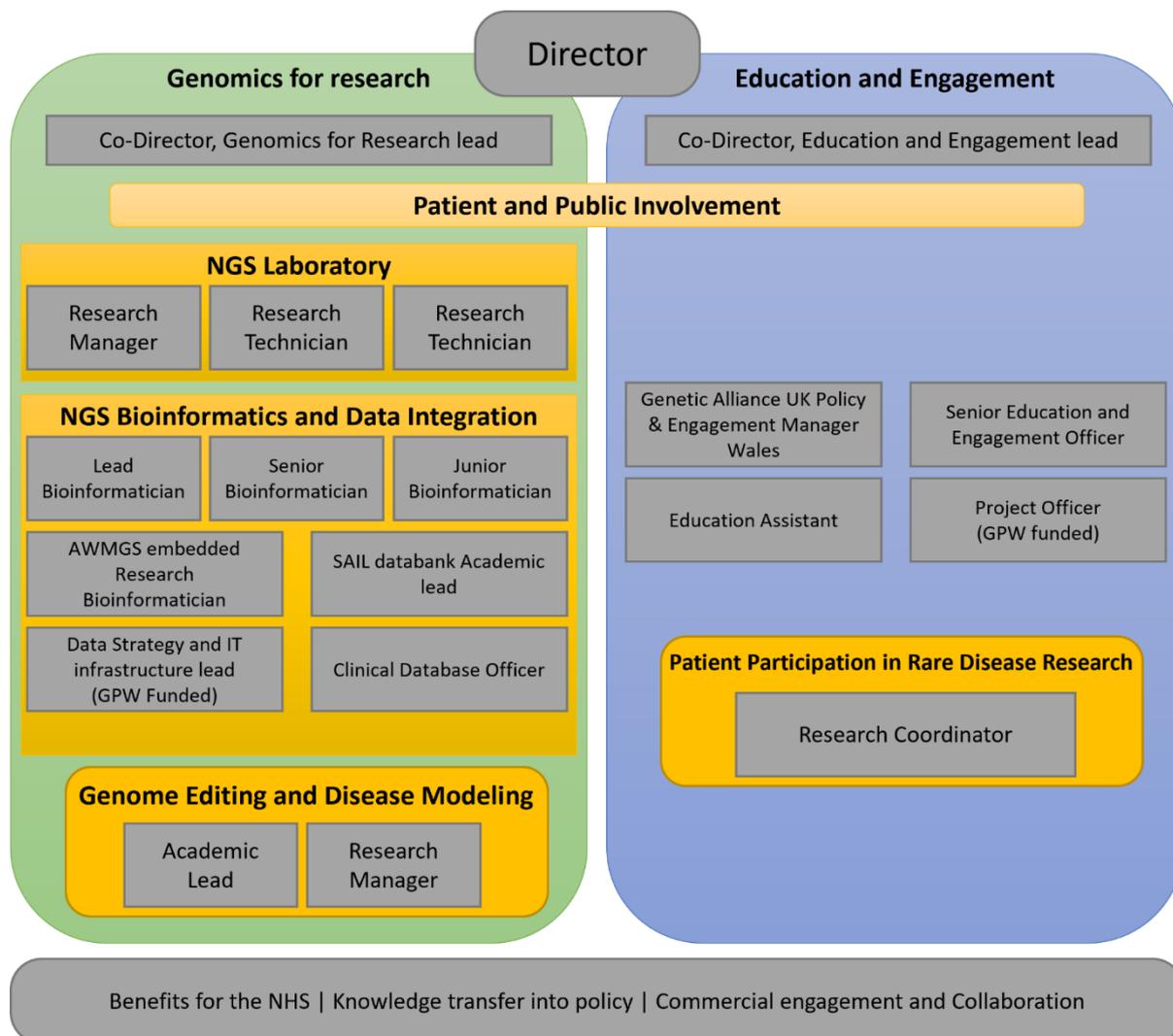
Strategic Objectives

The Wales Gene Park Objectives are to provide and develop expertise and infrastructure that will help Wales compete at the forefront of genetic and genomic research in its areas of priority and strength, particularly by supporting Health and Care Research Wales funded research activities and programmes. We will do this through providing support for Welsh researchers working collaboratively to help drive the development of Genomic Medicine in Wales. In doing so Wales Gene Park works to provide leadership in rare genetic disease research to help Wales to develop capacity in genomics, to undertake genomic analysis on a significant scale and support the linkage of genomic information to clinical data and other relevant data sets. These activities are underpinned by the objective of raising awareness and understanding, to enhance public and professional awareness and understanding of health-related genetic and genomic research and the opportunities and challenges this research brings.

RESOURCES AND WGP STRUCTURE

The Wales Gene Park annual budget employs a team of staff who are supported by senior academics, experts from the National Health Service (NHS) in Wales, managers and administrative staff from the host institution, Cardiff University.

Wales Gene Park Structure 1st April 2022- 31st March 2023.



More information about the teams is available on the Wales Gene Park website:

<https://www.walesgenepark.cardiff.ac.uk/our-teams/>

WHO'S WHO & KEY PARTNERS

Academic Partners

- Cardiff University
- Swansea University
- Bangor University
- Aberystwyth University
- Glyndwr University
- University of South Wales
- Cardiff Metropolitan University

NHS Partners

- Cardiff and Vale University Health Board
- All Wales Medical Genomics Service
- Genomics Partnership Wales

Commercial Partners

- Agilent Technologies
- Cellesce Ltd.®
- GW Pharmaceuticals Plc
- Illumina
- Illumine Theatre
- New England BioLabs, (UK) Ltd
- PTC Therapeutics
- Theatr Genedlaethol

Third Sector Funders and Partners

- Bowel Cancer West
- Bowel Cancer Wales
- Cancer Research UK
- Cancer Research Wales
- Genetic Alliance UK
- Pathological Society (GB & Ireland)
- Rare Disease UK
- SWAN UK
- Techniquest
- Tenovus Cancer Care
- The Wellcome Trust
- Tuberous Sclerosis Alliance (USA)
- Tuberous Sclerosis Association (UK)

Executive Management Team

- Dr Andrew Fry, Director
- Dr Karen Reed, Co-director, Operations Manager, Genomics for Research Lead (until Dec 2022)
- Mrs Angela Burgess, Co-director, Education and Engagement Lead
- Ms Sherrie Witts, Finance Manager
- Dr Hywel Williams, Impact Lead
- Dr Kevin Ashelford, Data strategy and IT infrastructure Lead
- Professor Nick Allen, Genome Editing Lead
- Professor Kerina Jones – Data linkage Lead
- Mrs Emma Hughes, Policy and Engagement Manager (Wales) Genetic Alliance UK, PPI Lead
- Professor Andy Tee, Commercial Interaction Lead

Patient Representatives

- Mr Alan Thomas
- Mr Nathan Davies

Strategic Advisory Group (SAG)

- Chair: Professor Colin Dayan, School of Medicine, Cardiff University
- Mrs Emma Hughes, Third Sector Representative and WGP PPI Lead
- Dr Rob Orford, CSO Health, Welsh Government
- Dr Dee Ripley, Deputy Chief Scientific Adviser for Health Welsh Government
- Chris Newbrook, Head of Health Sciences Branch, Welsh Government
- Dr Claire Morgan, Programme Director for the MSc Genomic Medicine, Swansea University
- Dr Ramsey McFarlane, Bangor University
- Professor Richard Adams, Director of Cancer Trials, Cardiff University
- Dr Mick Hunter, Entrepreneur in Residence Evotec, COO Viatem Ltd, and CEO Orca Pharmaceuticals Ltd
- Joanne Ferris, Operations Manager, the Association of the British Pharmaceutical Industry
- Clive Morgan, Managing Director, All Wales Medical Genomics Service, Cardiff and Vale University Health Board
- Sian Morgan, Head of NHS All Wales Medical Genomics Service Laboratory
- Michaela John, Head of Programme, Genomics Partnership Wales
- Dr Mark Bale, Head of Science Partnerships, Genomics England
- Dr Francis Sansbury, Health and Care Research Wales Specialist Lead for Genomics and Rare Diseases

LAY SUMMARY

Wales Gene Park is funded by Welsh Government through Health and Care Research Wales to support research into genomics (the study of an organism's genome – its genetic material – and how that information is applied). During the 2022-2023 reporting period, we supported research via two areas:

Area 1 – Genomics for Research

- Sequencing (determining the order of a DNA fragment) and analysing genomic information to support researchers.
- Making and using pre-clinical models of disease through genome editing (inserting, deleting, modifying or replacing DNA in the genome of a living organism) and other methods.
- Developing IT infrastructure that enables research using genomic data.

Area 2 – Education and Engagement

- Enhancing awareness and understanding of health-related genetic and genomic research for the public and health professionals.
- Involving those with lived experience in research prioritisation, development and delivery.
- Influencing Welsh Government policy development.

Summary of Activities

Sequencing Provision

Next Generation Sequencing (NGS) is a powerful, cost-effective, time-efficient way to sequence genes and genomes. At Wales Gene Park we use sequencing machines within Cardiff University and the NHS, All Wales Medical Genomics Services (AWMGS) to generate sequence data for researchers. We have enabled sequencing access for 68 projects, including areas such as cancer research, immunology and rare diseases.

Establishing the IT Infrastructure for Genomic Research

We work with academic (Universities), healthcare (the NHS) and industrial partners to improve the computational and data storage resources essential for genomics research. We continue to provide dedicated IT infrastructure to support genomic research through the Sêr Cymru IT infrastructure and the planned infrastructure at WGP's new home at Cardiff Edge.

Bioinformatic Analysis

Bioinformatics is the way we interpret complex biological data, such as genomic sequence data, using computers. At Wales Gene Park, our team of bioinformaticians (people who interpret the data) have supported 67 research projects this year and provided access to computing capacity to a further 38 researchers and postgraduate students. Wales Gene Park is now looking after over 384 terabytes of sequencing data held on behalf of researchers across Wales.

Genomic Data Integration into the SAIL Databank

This project looks at ways to put genomic data into the SAIL Databank. We have now installed a pipeline to process genetic data within a secure research environment. The plan is that during this processing any identifiable personal information would be removed and annotated files would be suitable for upload into the SAIL Databank following the normal process.

Developing Models of Disease

At Wales Gene Park we use cutting-edge methods (such as using specialised stem cells, CRISPR genome editing, and a range of cell and 'mini-organ' technologies) to generate new human genetic models of disease to undertake pre-clinical research.

We have supported 15 research groups on 18 projects, supporting 10 new grant proposals ranging from PhD studentships and early career fellowships to UK Research and Innovation (UKRI) project grants and drug discovery initiatives. Projects have looked at conditions including Alzheimer's disease, Huntington's disease, Glioma, and Motor Neuron Disease.

Rare Disease Research

The Inherited Tumour Syndrome Research (ITSR) group work on several different rare disease studies, funded by different organisations. Professor Julian Sampson is the Chief Investigator for *Genes and the kidney in Tuberous Sclerosis*, a study funded by WGP and the Tuberous Sclerosis Association, which aims to investigate renal disease in patients with tuberous sclerosis. The study has recruited 288 patients and is due to close in August 2023.

Researcher Dr Hannah West is the Chief Investigator for our *Genetic mechanisms in polyposis of the bowel* study which aims to discover novel genetic mechanisms underlying polyposis of the bowel and the development of tumours in this group of disorders. This study, funded by Ser Cymru and WGP, has reached its recruitment target of 375 patients and is now in follow-up.

Dr Laura Thomas is Chief Investigator for two of our studies. The *Molecular genetic analysis of duodenal polyposis in the inherited colorectal adenoma and cancer predisposition syndromes* study is investigating if patients with familial adenomatous polyposis (FAP) and MUTYH associated polyposis (MAP) are also at risk of developing tumours in the duodenum as well as the colorectum. This study has been funded by a number of organisations including WGP, Health and Care Research Wales, WCRC, Accelerate, Bowel Cancer West and Swansea University and has recruited 69 patients.

Dr Thomas' other study, *Exploring genetic causes of duodenal polyposis using healthy volunteers*, is funded by Accelerate, Swansea University and WGP, and explores the genetic causes of duodenal polyposis by comparing affected patients with healthy volunteers.

Our researchers (including a Research Coordinator funded through WGP) are also working on the first multi-centre European study of duodenal disease in MAP that aims to provide evidence as to whether surveillance recommendations developed for patients with FAP are also appropriate for patients with MAP. This study has recruited 650 patients with MAP and will continue for 20 years in total.

Education and Engagement

The Education and Engagement team have delivered a large programme of events and activities to raise awareness and increase understanding of the advances of genomics in Wales. Current audiences include health care professionals, patients and families, schools and colleges, and the public. We also prioritise involvement of the public and patients, using our networks to ensure their views are communicated clearly and effectively to Welsh Government. This year, through events organised by Wales Gene Park, we have reached:

- 1136 Health Professionals: 20 events
- 269 Schools/Colleges students/teachers: 5 events
- 2463 Public/Patients: 56 events

There are a huge range of activities which enable the team to raise awareness of genomics including, community talks, public lectures, genomics cafes and conferences. Following award success for their audio drama *Tremolo* last year, a new bilingual audio drama was developed by Illumine Theatre and Genomics Partnership Wales; *Deuce* which highlights the inherited condition, hypertrophic cardiomyopathy.

Case Study 3 shines a spotlight on the *Genomics After Dark* event held in Techniquest.

Enabling patient involvement in research

Wales Gene Park continues to facilitate opportunities for patient and public involvement in genomic research. Opportunities are circulated to our rare disease patient network, the Genomics Partnership Wales Patient and Public Sounding Board and through Genetic Alliance UK's Member Newsletter which is circulated to our 220 member organisations on a weekly basis. Attendees at events such as the Genomics Cafes were also made aware of opportunities for involvement. In total, 23 involvement opportunities were advertised with 166 individuals taking them up during the reporting period.

Examples of how we have involved people in projects focussed on genetics and genomics:

- Workshop with Cystic Fibrosis Trust and GPW Patient and Public Sounding Board to inform research where attendees fed back on the project objectives.
- Input to Medical Research Council (MRC) Rare Disease Research Node bids – supported bids led by Prof William Griffiths at Swansea University and Dr Jamie Duckers from Cardiff & Vale - support from PPI perspective
- Consultation with Patient Empowerment Group (PEG) to improve collaboration between rare disease registries across UK and provide feedback on the Welsh registries expansion to a service registering adults with rare diseases
- Bioethics consultation with Nuffield Trust – members of the Sounding Board attended this event to feed in from the patient perspective

Case Study 2 highlights the importance of working in partnership with patients and patient organisations to ensure the patient voice informs the development of service provision in the NHS.

CORE METRICS

Reporting period 1st April 2022 – 31st March 2023

**Health and Care Research
Wales infrastructure
award to the group**



Direct
funding
awarded
£734K

Jobs created
through direct
funding



Grants won during reporting period

Grants won	Led by group	Group collaborating
Number	2	10
Value	£108K	£3.53M
Funding to Wales	£108K	£3.42M
Funding to group	£108K	£142K
Additional jobs created for Wales	3	10.4
Additional jobs created for group	3	0.7



Number of publications



Number of public engagement
events



Number of public
involvement opportunities

WORK PACKAGE 1

Genomics for Research

Core Activities

- NGS for a range of applications using DNA and RNA
- Bespoke bioinformatic analysis for novel sequencing applications
- Pipeline analysis of standard data outputs (genomes, exomes etc.)
- Computing, processing and data storage for genomic medicine research
- Pathway development for the integration of genomic data into Secure Anonymised Information Linkage (SAIL) databank (Swansea)
- Contribution to healthcare professional education
- Derivation and growing human iPS (Induced Pluripotent Stem Cells) cells
- Utilising CRISPR (Clustered Regularly Interspersed Short Palindromic Repeats) genome editing technology to generate new models of disease for research
- Consultation for researchers requiring support in cell and genome editing techniques

Purpose and Progress

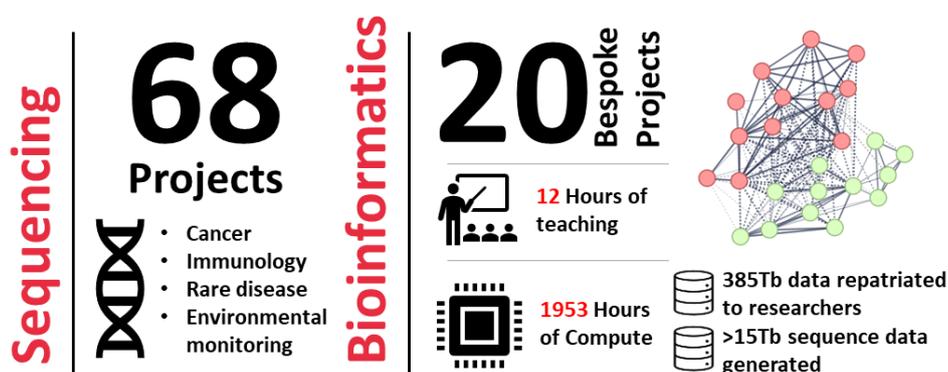
Wales Gene Park supports and promotes the application of genomic technologies via a range of activities delivered through:

- An NGS laboratory (68 projects supported in the last year)
- Bioinformatics provision (20 projects this year)
- Data science/integration expertise
- Genome editing and disease modelling.

Innovation is supported through collaborations with academia, the NHS, AWMGS, Pharma and Biotech sectors.

The provision of access for flexible NGS for research, together with expert bioinformatics support underpinned by dedicated high-performance computer infrastructure for genomic analysis, ensures the quality and quantity of genomic research undertaken in Wales and sees a greater proportion of externally awarded research funding is spent in Wales.

Wales Gene Park Genomics Facility Activity



Sequencing provision

NGS is a powerful, cost-effective, time-efficient technique used to sequence genes and genomes. At Wales Gene Park, the genomics facility lab team of 3 individuals use sequencing machines within Cardiff University and the AWMGS to generate sequence data for researchers. This approach allows a cost-effective strategy, optimising capacity utilisation to maximise the use of the sequencing machines. We have enabled sequencing access for 68 projects researching areas of cancer biology, rare disease research, arthritis, kidney disease, immunological conditions, neurodegeneration, and Alzheimer's projects. Additionally, WGP is facilitating sequencing for a Wastewater Monitoring Programme studying anti-microbial research (see Case Study 4). This approach for sequencing sees a greater proportion of externally awarded research funding is spent in Wales, contributing to the costs of the service maintenance for the sequencers (e.g., the NovaSeqs within AWMGS), and provides researchers with excellent quality data in a timely way.

Establishing the IT infrastructure for genomic research

We work with academic, healthcare and industrial partners to improve the computational and data storage resources essential for modern genomics research. We continue to provide dedicated IT infrastructure to support genomic research through the Sêr Cymru IT infrastructure and the planned infrastructure at WGP's new home at Cardiff Edge.

Bioinformatic analysis

Bioinformatics is the science of analysing complex biological data, such as genomic sequence data, using computers. At Wales Gene Park, our team of four bioinformaticians have supported 67 research projects this year and provided access to computing capacity to a further 38 researchers and postgraduate students. Wales Gene Park is now custodian to over 384 terabytes of sequencing data held on behalf of researchers across Wales.

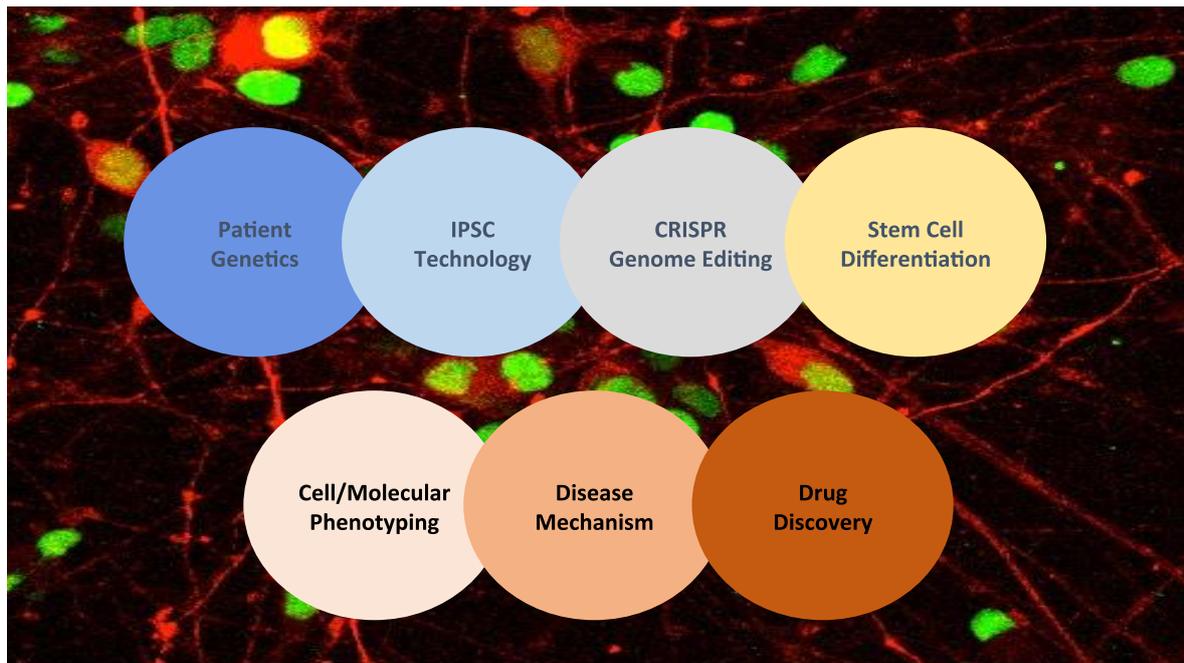
Genomic data integration into the SAIL Databank

Funded by Health and Care Research Wales, this is a pathfinder project being undertaken by PhD student Rob Maddison, looking at the methods of integrating genomic data into the SAIL Databank. We have previously described how files holding genetic variant information for individuals who donated samples to the Swansea Neurology Biobank were uploaded and annotated within SAIL. The annotation process identifies rare and potentially damaging variants, and we were able to investigate the presence of such variants in groups of individuals with different epilepsy outcomes. This was possible by linking the annotated variant dataset to routinely collected data from GP and hospital records and with the information derived from clinic letters. The results of this work were presented at the International League Against Epilepsy (ILAE) European Epilepsy Congress in July 2022 and at the ILAE meeting in Cardiff in October 2022. They have also been submitted for publication in a peer-reviewed journal.

The challenges identified by this study informed the next stage of the project and led to the installation of a pipeline to process genetic data on a separate server within a secure research platform (SeRP). The intention is that during this processing any identifiable personal information would be removed and the annotated files would be suitable for upload into the SAIL Databank following the standard procedure. The separate pipeline also allows for the annotation software to be updated so that the genetic datasets (Variant Call Format [VCF] files) can be reannotated with the latest datasets.

Developing Models of Disease

Advances in genomics are having unprecedented impact on our understanding of the genetic basis of disease, from the identification of mutations responsible for rare genetic disorders to understanding the complex interactions of multiple (polygenic) genes that increase a person risk of developing common disorders such as dementia or some cancers. Pathways to therapy require us to understand gene functions, firstly in models of disease. At Wales Gene Park we use cutting edge induced pluripotent stem cells (iPSC), CRISPR genome editing, cell differentiation and organoid technologies to generate new human genetic models of disease to undertake pre-clinical.



We have supported 15 research groups on 18 projects, supporting 10 new grant proposals ranging from PhD studentships and early career fellowships to UK Research and Innovation (UKRI) project grants and drug discovery initiatives. Projects have addressed Alzheimer's disease, Huntington's disease, Glioma, and Motor Neuron Disease. The figure illustrates our integration of genetic and stem cell technologies to understand the genetic bases of disease and establish platforms for drug discovery translational research.

Rare Disease Research

The Inherited Tumour Syndrome Research (ITSR) group work on several different rare disease studies. Professor Julian Sampson is the Chief Investigator for the *Genes and the Kidney in Tuberous Sclerosis* study (funded by WGP and the Tuberous Sclerosis Association) which aims to determine the natural history of renal disease in patients with the TSC2/PKD1 contiguous gene deletion and compare this with patients with mutations in TSC2 or TSC1 alone. The study has recruited 288 patients and is due to close in August 2023.

Researcher Dr Hannah West is the Chief Investigator for our *Genetic Mechanisms in Polyposis of the Bowel* study (funded by Ser Cymru and WGP) which aims to discover novel genetic mechanisms underlying polyposis of the bowel and the development of tumours in this group of disorders. This study has underpinned the identification of a genetic change that reduces the activity of a known tumour suppressor gene, causing the polyposis phenotype seen in a 4-generation family. This genetic

change was not identified from the standard clinical diagnostic services because it does not occur in the main body of the gene. However, this highlights the potential usefulness of expanding the diagnostic screen, particularly for patients with suspected polyposis when a classical genetic change has not been identified. This study has reached its recruitment target of 375 patients and is now in follow-up.

Dr Laura Thomas is Chief Investigator for two of our studies. The *Molecular Genetic Analysis of Duodenal Polyposis in the Inherited Colorectal Adenoma and Cancer Predisposition Syndromes* study (funded by a number of organisations including WGP, Health and Care Research Wales, WCRC, Accelerate, Bowel Cancer West and Swansea University) is investigating if patients with familial adenomatous polyposis (FAP) and MUTYH associated polyposis (MAP) are also at risk of developing premalignant and malignant tumours in the duodenum as well as the colorectum. This study investigates the genetic factors, inherited and somatic, associated with growth and progression of duodenal adenomas to cancer in MAP and has recruited 69 patients.

Dr Thomas' other study is *Exploring Genetic Causes of Duodenal Polyposis Using Healthy Volunteers* (funded by Accelerate, Swansea University and WGP) which uses 3D organoid models to explore the genetic causes of duodenal polyposis by comparing affected patients with healthy volunteers. A comparison of healthy volunteers with 3D duodenal organoids established from patients with FAP and MAP (established as part of the *Molecular Genetic Analysis of Duodenal Polyposis in the Inherited Colorectal Adenoma and Cancer Predisposition Syndromes* study) can help to determine how polyps are arising in patients with these conditions and we have recruited 13 patients from Cwm Taf Morgannwg University Health Board.

Our researchers (including a Research Coordinator funded through WGP) are also working on the first multi-centre European prospective study of duodenal disease in MAP that aims to provide evidence as to whether surveillance recommendations developed for patients with FAP are also appropriate for patients with MAP. It aims to collect long-term data on the endoscopic findings and provide follow-up information to aid understanding of the natural history of duodenal disease in MAP, taking into account that some patients may require therapeutic procedures including removal of polyps where there is advanced duodenal disease. It will also prospectively collect data on the occurrence of colorectal cancer and extra-intestinal cancers. This study has recruited 650 patients with MAP and will continue for 20 years in total.

Publications and presentations:

- Dominguez-Valentin M et al. *Mortality by age, gene and gender in carriers of pathogenic mismatch repair gene variants receiving surveillance for early cancer diagnosis and treatment: a report from the prospective Lynch syndrome database* EClinicalMedicine. 2023 Mar 20;58:101909. doi: 10.1016/j.eclinm.2023.101909.
- *Inherited MUTYH mutations cause elevated somatic mutation rates and distinctive mutational signatures in normal human cells*. Robinson, P., Thomas, L et. Al Nat Commun. 2022 Jul 8;13(1):3949. doi: 10.1038/s41467-022-31341-0
- Prof. Julian Sampson gave a talk to the UK Cancer Genetics Group spring meeting in Leeds on *Targeted Treatment for Tuberous Sclerosis: Benefits and Limitations of mTOR Inhibitors in Clinical Practice* which is based on previous WGP supported work.
- PhD student Becky Truscott was a speaker at the European Hereditary Tumour Group annual conference. Her talk was called *Duodenal Disease in 579 Individuals with MUTYH-associated Polyposis: Updated Findings from an International Prospective Observational Study*.
- PhD students Becky Truscott and Angharad Walters gave a talk on their research at the Public Genomics Café with Genomics Partnership Wales in April 2022

WORK PACKAGE 2

Education & Engagement

Purpose

Wales Gene Park carries out a wide portfolio of activities in professional and public education and engagement. The programme provides opportunities for engagement and involvement of patients and families in research, service development and health and social care policy. Wales Gene Park's education activities draw on expertise in the Welsh Higher Education Institutions and NHS, and on the wider UK and international genomics community.

Core Activities

- Education and engagement events and activities via public and schools' programmes
- Support through events, initiatives, and campaigns, for those affected by Rare Diseases
- Management of networks for each of WGP's four key stakeholder groups (patients, schools, the public and professionals)
- Recruitment to Health and Care Research Wales and NIHR genetics clinical research studies
- Approved continuing professional development (CPD) in genetics and genomics for those working in healthcare and allied professions
- Public input to Welsh Government policy in genetics and genomics, including rare diseases

Progress

Public

A varied and wide-ranging programme of engagement activities were held for public groups which included:

Genetics & Genomics for the 3rd Generation (3G) Public Conferences

Now in its 7th consecutive year, the annual 3G conference took place virtually in December. The event is aimed at the over 50 age group, but open to any members of the public with an interest in DNA, genetics, genomics and related topics. Over 100 attendees joined via Zoom to enjoy talks including *The Brave New World of CRISPR Gene Editing*, *Exploring Early Human Populations in Britain Using Ancient DNA*, *Ethical Dilemmas in Genetic/Genomic Medicine* and *Moving Through the Menopause*.

- A superb event! Impressive as ever, much appreciated *3G attendee*
- Thank you for a fascinating day. I learnt so much *3G attendee*



Community Group Talks

Thirty-three DNA and genetics/genomics-related talks were given to community-based organisations including The Rotary Club, Women's Institute groups, Probus Clubs, The University of the Third Age, retirement fellowships, library groups and social clubs throughout Wales. In-person presentation resumed following the pandemic, and topics included Genetics and Genomics in Everyday



Life, Epigenetics, Genomics in the Media, Personalised Medicine, and DNA Fingerprinting.

Genomics Cafes

Public Genomics Cafés form part of the Genomics Partnership Wales engagement programme that WGP designs and delivers. Free and open to all, the cafes include short, informal talks about genomics relating to health and medicine with guest slots from health professionals, researchers, third sector groups and those affected by genetic and rare conditions, who share personal experiences. Since April 2022, five cafes have taken place for the general public, attended by more than 275 people. Examples of talks included *Mini-guts for Cancer Research*, *Enabling Diagnosis through Genomic Research*, *A Personal Account of Thalassaemia*, *Genetics & Exercise*, *Supporting those with Undiagnosed Conditions in Wales: the new SWAN Clinics Initiative*, *Genomic Data-Sharing for Research*, *A Family's Experience of the Rare Condition Neurofibromatosis type 1*.

- Thank you all, from a first-time attendee, looking forward to joining again! *Public Genomics Café attendee*
- What a privilege it is to hear these stories and experiences today. Thank you. *Public Genomics Café attendee*
- Thoroughly absorbing morning! Thank you very much to all contributors and for clear presentations that could be followed by non-specialists like me! *Public Genomics Café attendee*



Following the positive response to the Public Cafés, separate Young People’s Genomics Cafés were initiated. Aimed at those aged 16 to 25, they are held three times a year on a termly basis, and in the last 12 months were attended by over 140 people. Topics included *What is a Genome and what can we do with it?*, *Chromosomes!*, *An Introduction to Pharmacogenomics*, *Genomics Careers Pathways in the NHS: Where Could They Lead You?*

- Thank you! I found the session about pharmacogenomics interesting in particular. *Young People’s Café attendee*
- Thank you to all the speakers and host, this was incredibly interesting and eye opening! *Young People’s Café attendee*



A blend of online and in-person cafes will return over the coming months and there are also plans for a ‘Café Roadshow’ throughout Wales.

Genomics After Dark @ Techniquet

As part of an ongoing programme of communication and engagement with the public around genetics/genomics, the inaugural *Genomics After Dark* outreach event was organised and delivered by Wales Gene Park’s Education and Engagement Team in conjunction with Genomics Partnership Wales. Held at the Techniquet Science Discovery Centre in Cardiff, the event was aimed at an adult audience (particularly the hard-to-reach 18 to 50 age demographic) and free to attend. It comprised a range of engaging, hands-on activities which included:

- Laboratory-based DNA extraction workshops
- Public talks focusing on ethics in clinical genetics/genomics
- Virtual Reality games created by WGP and GPW
- Careers videos highlighting job roles
- A wide range of interactive stands from organisations across sectors such as healthcare, research, public health, higher education and the arts



Further details of *Genomics After Dark* can be found in Case Study 3.

Public Lectures

Wales Gene Park's public lectures are aimed at engaging with a lay audience about genetics and genomics-related subjects. Attendees are varied and range from Sixth Form students through to retired members of the public. An example from year's programme was an excellent lecture on Pharmacogenomics from Professor Dyfrig Hughes, Bangor University, held at Pontio, Bangor.



Can genetic testing help make medicines safer?

Professor Dyfrig Hughes, Bangor University

Medicines have transformed the lives of many. However, there is always a chance that people experience a side effect. These can range in severity from

Genomics Partnership Wales

Large Lecture Theatre, Level 5, Pontio, Bangor University, Deiniol Road, Bangor LL57 2TQ

Monday 6th March 2023, 7 - 8pm (registration from 6.30pm, talk starts 7pm)



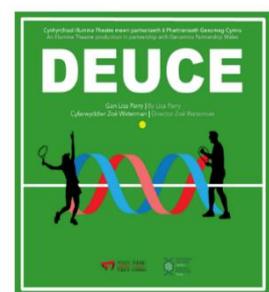
- Wales Gene Park also participated in a range of other public events, highlights of which include:
- Hands-on *Discovering DNA* stands at the summer and autumn Integrated Wellbeing Network Gwent (Aneurin Bevan UHB) Cwtsh Festivals in the Rhymey Valley.
- A two-day interactive stand at Pontio, Bangor
- A genetics/genomics information stand in the Science and Technology Village at the National Eisteddfod of Wales, Tregaron



Deuce: a new bilingual audio drama from Illumine Theatre and Genomics Partnership Wales

Produced by [Illumine Theatre](#) in partnership with Genomics Partnership Wales and in conjunction with Wales Gene Park, *Deuce* is a podcast drama written by Cardiff-based playwright Lisa Parry and directed by Zoë Waterman, which explores hypertrophic cardiomyopathy (HCM). Translated by Branwen Davies, there will be a Welsh and English version available to stream for free, to ensure a wide range of listeners can experience this powerful and thought-provoking production. The 30-minute podcast tells the story of Alys, a teenage tennis player who collapses during the final of the Girls' Junior Championship at Wimbledon. As first aiders realise she is in cardiac arrest, she is rushed to hospital, where she lies in a coma. Here, she is visited by her late father, Daf, a previous professional tennis player who Alys hopes to emulate.

The world as Alys knows it is then turned upside down when doctors diagnose her with hypertrophic cardiomyopathy (HCM); a condition where the muscular wall of the heart becomes thickened and pumping blood around the body becomes more difficult. With renowned professional athletes like footballer Fabrice Muamba and basketballers Hank Gathers and Will Kimble famously diagnosed, the condition has frequently appeared in real-world headlines due to its potential impact. Not only does Alys understand that her diagnosis could spell the end of her own career, she realises that this inherited condition, caused by a fault in her genome, could have also been the cause of her father's so-far unexplained death.



Lisa's script, informed by the experience of experts in genetics and genomics, cardiologists and those living with hypertrophic cardiomyopathy, explores the life-changing impact of diagnosis on relationships, ambitions and mental health. As Alys leans on her father for support in coming to terms with the idea of returning to her body and to a life without professional tennis, she begins to understand that she must ultimately move beyond the grief she feels for him, while navigating a challenging new reality.



“Researching this drama was an incredibly moving experience where people shared their stories. It feels right to tell this through audio, which is a really intimate form of storytelling where the listener conjures the images in their own mind. I love the fact people will be able to listen to it on their phones, wherever they are, and can't wait to get into the rehearsal room.” *Lisa Parry, playwright*

“Following the success of TREMOLO, we know that this creative approach helps the wider population understand the relevance and importance of genomics for individuals and families. This will be key as technology improves, and our understanding and application of genomics increases.” *Michaela John, Head of Programmes for Genomics Partnership Wales (GPW).*

DEUCE will be launched in summer 2023 and be free to access via platforms including Spotify and Apple Podcasts.

Award success for Tremolo – an audio drama about early-onset familial Alzheimer’s disease for post-16 students and the wider public

Tremolo by Lisa Parry - an exciting, collaborative project between Wales Gene Park, [Illumine Theatre](#) and [Theatre Genedlaethol Cymru](#) - is a thought-provoking, bilingual podcast drama exploring the impacts of a diagnosis of early-onset Familial Alzheimer’s Disease (eFAD), and some of the socio-ethical issues around genetic testing.



Following its launch in March 2022, *Tremolo* (see Case Study in Wales Gene Park’s 2021-22 Annual Report) has received BBC Audio Drama nominations and award success! Actor Gareth Elis won the *Marc Beeby Award for Best Debut*



Performance at the recent [BBC Audio Drama Awards 2023](#), announced at a ceremony in the Radio Theatre, BBC Broadcasting House in March 2023. Gareth received the award for his portrayal of 18-year-old Harri who’s just finished his exams and is ready to follow his dreams when he discovers that his mother has been diagnosed with the condition, which he and his sister each have a 50 per cent chance of inheriting. A nomination for *Tremolo* also came in the Best Original Single Drama category.

Read more about *Tremolo*, and the accompanying Education Park for schools and colleges, [here](#). *Tremolo* is available in both English and Welsh to listen and download via platforms including [Spotify](#), [Apple Podcasts](#) and [AM](#).



Public Genetics Network

Wales Gene Park also continued to engage with members of the public through its *Public Genetics Network* and associated biannual newsletter which provides news, information and details of events and opportunities.

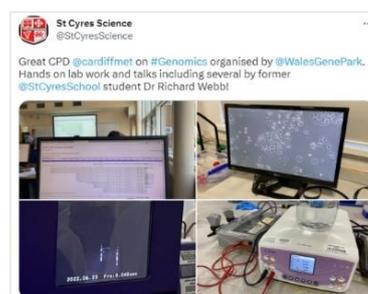
Schools and Colleges

During the last 12 months, Wales Gene Park's engagement activities for post-16 School and Colleges have gradually started to recover from the severe disruption of the Covid-19 pandemic, although the impact of this is still felt over the last year. Despite this, Wales Gene Park delivered a varied programme of engagement which included:

- Engagement via Wales Gene Park's **Teachers' Genetics Network** and associated termly newsletters
- Genetics/genomics-focused careers stands at school and colleges, and work placements hosted by Wales Gene Park
- Interactive information stands at events, highlights of which included a Women in Science Technology Engineering & Maths (STEM) events at Techniquet, and the *Life Sciences Challenge* and *Science in Health* Sixth Form event at Cardiff University's School of Medicine
- Bespoke genetics/genomics education at schools, as well as a session on Ethical Dilemmas in Clinical Genetics in partnership with the All-Wales Medical Genomics Service as part of Cardiff University Sutton Trust Summer School
 - *One of my most favourite sessions, so thought provoking Student, Sutton Trust Summer School attendee*



- A 'Cutting-Edge Biology' Genetics & Genomics CPD event for secondary biology teachers, held in collaboration with Techniquet, Cardiff Metropolitan University and STEM Learning. The day-long course provided the 40 attendees with updates on research and advances in the fast-moving field of genetics and genomics and included talks from expert speakers on topics including Nutrigenomics, Analysing COVID-19 Using Genomics in Wales, and Careers in Biosciences. There was also a hands-on bioinformatics workshop on Identifying Genomic Variants Associated with the Genetics of Taste, as well as a corresponding laboratory practical session with follow-up discussion/debate on genetics & ethics including sequencing genomes, and consent.
 - *Excellent hands-on experience! Attendee, Teachers' CPD*
 - *Thanks for organising - really useful and enjoyable Attendee, Teachers' CPD*



Health Professionals and Researchers

Throughout the year a range of activities were facilitated for health professionals and researchers working in the field of genetics and genomics, which included:

Specialty Certificate Examinations (SCE) Revision course

The annual Clinical Genetics Revision Course, hosted by the All-Wales Medical Genomics Service, was held for doctors training and specialising in this area of medicine.

- The course was brilliant – Thank you! Thank you to everyone who was involved in organising and delivering it *SCE Course attendee*

Nanostring Spatial Biology Seminar

Wales Gene Park supported the organisation of this event hosted by Dr Hywel Williams, Division of Cancer Genetics, Cardiff University in partnership with Nanopore. The hybrid seminar explored Spatial Biology, an expanding collection of methods to examine biological molecules in their geographical context, and how it can accelerate research.



Peter Harper Memorial Symposium: Celebrating a Life in Medical Genetics

In September 2022, the genetics community came together to remember its late friend and colleague Professor Sir Peter Harper. The Memorial Symposium in Cardiff, organised by Wales Gene Park's Education & Engagement Team, brought together colleagues, from the UK and abroad, and members of Peter's family for a special day to celebrate his life and achievements, and his unique and lasting approach to the development of medical genetics that has had major impact locally, nationally and internationally.



In-person and virtual Bioethics workshops: Ethical Considerations in Genomic Healthcare and Research Initiatives in Wales

Wales Gene Park partnered with Genomics Partnership Wales and the All Wales Medical Genomics Service to facilitate two workshops to consider the ethical issues raised by genomic healthcare and research initiatives in Wales. The outcome of the workshop discussions fed into a national workshop and project hosted by the Nuffield Council on Bioethics entitled 'Realising the Ethical Commitments in the Genome UK strategy', which aims to develop a best practice approach to applying ethical standards in this area.

Cardiff Edge Staff Engagement

Over the last 12 months, activities have continued to increase around the forthcoming relocation of partners from across Genomics Partnership Wales - including Wales Gene Park, All Wales Medical Genomics Service, and the Public Health Wales Pathogen Genomics Unit (PenGU) - to the new Cardiff Edge site. A key aspect of this new co-localisation is to engage with, and involve, staff across all partners. As such, Wales Gene Park's Education & Engagement Team – working closely with GPW colleagues – has helped deliver a number of engagement events and activities, giving staff across all partner organisations an opportunity to meet, network, share information, ask questions and provide feedback and ideas. These events will continue up to, and beyond, the move.



Education sessions

A series of genetics/genomics-related sessions were delivered to support the education of professionals such as healthcare students, health professionals and researchers. These included:

- *Living with Genetic Conditions* sessions - which include personal narratives from individuals affected by genetic, rare or undiagnosed conditions - at higher education institutes throughout Wales, attended by nursing, midwifery and Allied Health Professional students. Sessions covered Fragile-X syndrome, tuberous sclerosis, cancer, and SWAN (Syndromes Without a Name)
- Sessions organised for Cardiff University Genomic Counselling MSc students which focused on patient/family/carer experiences
- Teaching sessions, incorporating lived experiences, to Cardiff University School of Medicine and School of Healthcare Sciences undergraduate and postgraduate students, as well as placements for Student Selected Components Medicine modules
- A continuation of Genomics Partnership Wales's *Hospital Genomics Roadshow* talks, delivered to health professionals across Wales, with the aim of raising awareness and informing audiences about advances in genomic medicine in Wales including the new *Genomics Delivery Plan for Wales 2022 – 2025*, launched December 2022.
- One aspect of my role as a lecturer within Cardiff University, School of Health Care Sciences, is to organise and develop the genetics/genomics education for the Bachelor of Midwifery (Hons) programme. Being able to liaise with the Wales Gene Park ensures that we can draw on the expertise of professionals working in this specialist service as well as service users. This combination of reliable, up to date information and application to practice is essential for evidence-based midwifery education and practice. It also highlights the benefits of partnership working - enabling individuals today to be the health care practitioners of tomorrow. *Janet Israel, Lecturer & Lead Admissions Tutor Bachelor of Midwifery (Hons) programme, Cardiff University School of Healthcare Sciences*



Genomics Information Stand

Wales Gene Park, also representing Genomics Partnership Wales, exhibited at numerous scientific and healthcare conferences, meetings and careers-focused events throughout the year, with information stands and hands-on activities. These included:

- Minority Ethnic Communities Health Fair, Cardiff
- Primary Care Training Days, including a CPD event at Morriston Hospital
- Cwm Taf Research & Development Conference
- Health Education and Improvement Wales QISTMas Conference
- Cardiff Metropolitan University Health Science Careers Fair
- Healthcare Science Cymru Conference
- Festival of Genomics, London



Supporting Research Engagement

Wales Gene Park's Education & Engagement team has continued to partner the 'Unique You - How your genome defines your healthcare' project led by researchers at Cardiff University. The project has involved working with public partners to co-design engaging, public-friendly resources – a game and an animation - to inform and raise awareness of genomics and precision medicine. Wales Gene Park is supporting and facilitating this research engagement project in several ways, including using its networks to connect researchers with community groups, supporting public engagement workshops, and helping to develop and trial new resources at its public events.

- Wales Gene Park's engagement team have been great at supporting our engagement project, through helping us advertise and recruit participants, providing constructive advice on the development of the project and enabling us to test the resources made at the Genomics After Dark event. *Dr Elaine Dunlop, Lecturer, Cardiff University*



Professionals' Genetics & Genomics Network

Wales Gene Park continued to engage with health professionals and researchers with an interest in genetics and genomics through its biannual *Professionals' Genetic Network* and associated newsletters, providing news, information and details of opportunities for education, research and more.

Patients & Families

The programme of activities to support and empower those affected by genetic, rare, and undiagnosed conditions included:

Events

Rare Disease Patient Network Annual Meeting

Wales Gene Park held its eighth annual Rare Disease Patient Network meeting in November 2022. The aim is to bring members of the Rare Disease Patient Network together as well as guest speakers and others with an interest in rare, genetic and undiagnosed conditions. The event featured talks including:

- Developing a patient passport service for supporting coordination in rare diseases
- How to get involved in raising awareness of rare diseases by taking part in activities for Rare Disease Day 2023
- Progress implementing new multi-disciplinary clinics for those affected by undiagnosed, rare conditions across Wales
- Update on implementation of the Welsh Rare Disease Action Plan in Wales

During the afternoon session, attendees were invited to take part in an online workshop focused on understanding priority areas for future rare disease research applications and developing a digital hub for rare diseases.

- Thanks so much for today, it's been so informative and insightful *Catriona - Child Growth Foundation*
- Really interesting discussions. Thanks for a great conference. Really inspiring and thought provoking. *Sabrina – All Wales Therapeutic Toxicology Centre*
- Thank you so much for letting me join the call. This has been so wonderful and inspiring! *Jill Harkin*

Rare Disease Day 2023 - highlights

- Wales Gene Park had a stand at Morriston Hospital and took along its Rare Disease Day selfie frame to raise awareness as well as taking part in a genomics training day with a large cohort of GP Trainees from across the Swansea Bay Health Board.
- Health and Care Research Wales publicised the benefit of rare disease research from a family perspective sharing Marie & Trystan's story of how research has benefitted TSC: [“We refused to think nothing could be done”, mum of son affected by rare disease highlights the importance of ongoing research | Health Care Research Wales \(healthandcareresearchwales.org\)](#)
- Wales Gene Park hosted a Young People's Genomics Cafe - one of the speakers, Daf Matheson, highlighted a documentary he made following his late uncle's experience of Cystic Fibrosis: [A Life With Less - Documentary Film - YouTube](#)
- Genetic Alliance UK in collaboration with Wales Gene Park organised two events to highlight the day - an in person parliamentary reception in Cardiff Bay and a joint virtual event in collaboration with the other 3 nations of the UK.

Rare Disease Day 2023 Parliamentary Reception

The annual Rare Disease Day parliamentary reception for 2023 in-person at the Norwegian Church, Cardiff Bay. Speakers included the Clinical Lead for Rare Diseases in Wales, Dr Jamie Duckers who spoke about implementing the Welsh Rare Disease Action Plan and Dr Graham Shortland OBE, Lead for the SWAN clinic in Wales. Genetic Alliance UK launched a report highlighting how well-coordinated care can make a real difference to quality of life for people living with rare conditions and their families, which was reinforced by SWAN parent speaker, Mandy Hughes at the event.



Rare Disease Day is an annual international event aimed at raising awareness and highlighting the needs of people with rare conditions. It provided an opportunity for the rare community to come together to raise awareness of the common issues affecting those living with rare conditions (see images).

- One of our Trustees was able to attend the Wales reception on the 14th Feb and I had a phone call with her today where she couldn't say enough positive things about the event! It had left her feeling empowered and inspired, so please pass on our thanks to all those involved. *Addison's Disease Self-Help Group*



Rare Disease Day Joint Nation Virtual Event

Alongside the in-person event, over 60 people attended an online joint-nation event. Over the past few years, the value of bringing the whole UK community together online, making new connections and reaching people who may not usually join our Rare Disease Day events, has been learnt.

Members from the four UK Nations shared the progress being made around care coordination, and there were also personal accounts from people from each nation who live with rare conditions.

From Wales, Lucy Dixon, Chair of PCD (Primary Ciliary Dyskinesia) Support UK, shared her experience of living with PCD and her experience of care coordination. Rhiannon Edwards, Coordinator of Rare Disease Implementation Group (RDIG) gave an update on Wales and its implementation of the Rare Disease Action Plan.

James Ingram, Director of Scienap gave an update about “Care & Respond,” the Health Passport that’s been developed with funding from the Welsh Government Health Hack and is now able to benefit patients with rare diseases.

- Thank you Louise and Team at Genetic Alliance UK and all the speakers today - wonderful to have the opportunity to join online *Marie James, Tuberos Sclerosis Complex and Rare Disease Advocate*
- Thank you so much for today's very informative and interesting event. Thanks to all the amazing speakers and Louise and all the staff at Genetic Alliance UK for all your clear hard work. *Karen Whitehead MBE*

SWAN UK Cymru events

Undiagnosed Childrens' Day, April: a Twitter take over was held on the Cymru channel and Amanda, the Engagement and Support Officer for Wales introduced her role and gave updates on Wales engagement. Theme was #PINKathon with pink hair styles and infographics and 'Twibbon' frames for members and partners to participate on social media.



SWAN Information Day, May: virtual SWAN UK information event with 61 people registered to attend, with a good attendance from Wales as a result of raised awareness on various networking events and social media platforms. A parent spoke about her experiences of having a child with an undiagnosed genetic condition.

SWAN Family Day, May: the first face-to-face family event for members in Wales took place with 12 adults and 14 children attending a bowling event, largely organised by our South Wales Parent Rep., and an opportunity to meet face to face with families.

SWAN UK Cymru Sounding Board, August: the Sounding Board consists of 6 SWAN UK members in Wales. They have come together for 2 virtual meetings and assisted with consultations regarding the SWAN Clinic and the roll-out of a pilot counselling project delivered by RareMinds, which will invite SWAN UK members in Wales to come together for a series of group counselling sessions. The aim is to create a safe and comfortable platform for the group to continue to support each other, peer-to-peer, after the block sessions.

SWAN UK CYMRU/Genetic Alliance UK presentation at Noah's Ark – August - a face-to-face meeting and presentation was given to inform staff at the Noah's Ark Children's Hospital at the University of Wales Hospital, Cardiff about SWAN UK Cymru and the work of Genetic Alliance UK in Wales.



Policy

Welsh Disease Action Plan - The SWAN Clinic & the 'Care & Respond' Patient Passport

WGP collaborated with the Welsh Government and Rare Disease Implementation Group to secure funding for the UK's first Syndromes Without a Name clinic in Wales. WGP has been coordinating input from the SWAN Cymru Council made up of parents of children affected by undiagnosed genetic conditions to help develop Patient Reported Outcome Measures (PROMs) and Patient Reported Experience Measures (PREMs) to measure the benefit to patients referred to the clinic. Monthly meetings are held with key stakeholders to provide updates that can be communicated to the SWAN community and a patient representative perspective is provided to support clinic developments and evaluation.

In December 2022, WGP organised an event bringing together key stakeholders of the SWAN clinic in Wales to improve communication



and engagement. A Parent Rep. and the Support Officer for SWAN UK Cymru joined to share a new Rare Resources information toolkit to support families.

Many Genetic Alliance UK member organisations have been actively involved in supporting development of a rare disease patient passport, 'Care and Respond' which can link with health care systems, including emergency care and social care. The passport was launched on Rare Disease Day 2023.

Rare Resources Cymru

Rare Resources Cymru was published in December 2022 and work began to disseminate the resources to key health and education professionals across Wales. The bilingual toolkit is a collection of information guides for families who have recently received a diagnosis of a genetic or rare condition, are on the journey to receive a diagnosis, or have been told their child's condition is so rare they may not get a diagnosis. The resources have been developed and produced by Genetic Alliance UK in collaboration with families in Wales and provide links to reliable sources of information and support and contain 'top tips' from families including information about how to find about and get involved in research. View the full list of resources here: [Rare Resources - Cymru | SWAN UK \(undiagnosed.org.uk\)](https://undiagnosed.org.uk)

Collaborating with Congenital Anomaly and Rare Disease Information Service (CARIS)

Working with the Welsh Registry for Rare Diseases to support development of their self-registration service for adults with rare diseases. Feedback has been provided on their self-referral forms and links made with member organisations to support piloting this work. There will be ongoing support for future plans for co-production to further develop the service and publicise the opportunity with member networks.

Rare Disease UK Patient Empowerment Group brought together the rare disease data registries from across the UK to discuss working collaboratively with our patient organisation network. CARIS contributed from the Welsh perspective.

Genomics Partnership Wales - Patient & Public Involvement (PPI) Sounding Board

The Genomics Delivery Plan for Wales was published in December 2022. WGP supported Genomics Partnership Wales to develop strong foundations 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 help strengthen the quality of genomics output.

WGP has been involved in working to establish the Genomics Sounding Board, made up of 30 people with diverse experiences of rare, genetic conditions and cancer testing or diagnosis. Working together, they have developed guiding principles that will serve as the foundation for future delivery; a commitment to communicate 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.

Throughout the 2022-3 reporting period, WGP offered 23 opportunities for people to get involved in projects, with 166 people taking these up. Examples of how we have involved people in projects focussed on genetics and genomics:

- Workshop with Cystic Fibrosis Trust and GPW Patient and Public Sounding Board to inform research where attendees fed back on the project objectives.
- Patient & Public Sounding Board – regular consultation with our Board of PPI members to provide feedback on developments and projects and to support evaluation of the programme

- Input to Medical Research Council (MRC) Rare Disease Research Node bids – supported bids led by Prof William Griffiths at Swansea University and Dr Jamie Duckers from Cardiff & Vale - support from PPI perspective
- Consultation with Patient Empowerment Group (PEG) to improve collaboration between rare disease registries across UK and provide feedback on the Welsh registries expansion to a service registering adults with rare diseases
- Bioethics consultation with Nuffield Trust – members of the Sounding Board attended this event to feed in from the patient perspective

ACHIEVING IMPACT

Impact in genetic and genomic research is of paramount importance as it holds the key to improving healthcare outcomes and transforming the lives of individuals and communities. Wales Gene Park's mission is to support impact-driven research from across Wales to bridge the gap between scientific discoveries and their real-world applications.

The activities of Wales Gene Park are firmly grounded in collaborative work spanning various healthcare specialties. The following examples highlight the remarkable impact that has been achieved through this cooperative way of working:

Case study 1

The Wales Gene Park is collaborating with its partners to create Trusted Research Environment (TRE) services for genomic data. These services will enable researchers to access genomic healthcare data in a safe and controlled manner and aim to advance the utility of patient data for research. Case Study 1 provides details of this initiative and showcases the work already undertaken to develop the information governance, data security, and IT infrastructure necessary for future TRE services.

Case study 2

Patient engagement is at the heart of Wales Gene Park's activities. Case study 2 demonstrates how their collaboration with charities has led to Lottery funding to establish SWAN UK Cymru, a Syndromes Without a Name network in Wales and how they have supported a bid to the Welsh Government to create a pilot SWAN clinic for two years.

Case study 3

With genetics and genomics becoming increasingly important in medicine and healthcare, public engagement and understanding are crucial. Case study 3 outlines the inaugural *Genomics After Dark* outreach event organised by Wales Gene Park's Education and Engagement Team at Techniquest. The success of this event was highlighted by the overwhelmingly positive feedback from attendees.

Case study 4

The rise of antimicrobial resistance represents one of the biggest global healthcare challenges. Case study 4 describes Wales Gene Park's involvement in a Welsh Government-sponsored project to monitor wastewater on a national scale covering 90% of the Welsh population. By evaluating the abundance of clinically relevant taxa, this project is consistent with global efforts to reduce the impact and spread of antimicrobial resistance.

CASE STUDY 1

Genomics Partnership Wales and Trusted Research Environment (TRE) Services

As Wales Gene Park we are working with our Genomics Partnership Wales partners — the All Wales Medical Genomics service, AWMGS, and Public Health Wales Pathogen Genomics service PenGU — to develop Trusted Research Environment services for genomic data that will allow researchers safe and controlled access to genomic healthcare data for healthcare research whilst protecting the interests and the privacy of Welsh patients.

Trusted Research Environments, or TREs, provide approved researchers with controlled access to health data to support their research. TREs are increasingly used by academia, healthcare, and industry as safe havens where appropriately vetted and trained researchers are granted controlled access to data under a strict governance regime that limits what can be done with the data and who has access. A well-designed TRE enables research whilst protecting the interests of the patients and public.

The TRE concept, therefore is an established route by which Wales can streamline researcher access to genomic clinical data and provide mechanisms for linking data with other healthcare and research data. A TRE has the potential to improve the usefulness of patient data for research data whilst freeing up valuable NHS staff time and, most importantly, protecting patients' privacy.

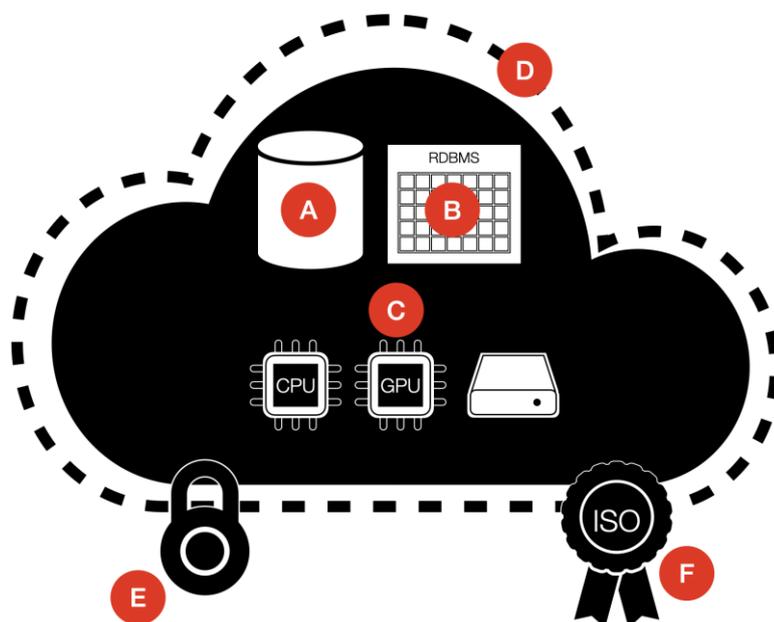


Figure 1: A Trusted Research Environment (TRE) provides approved researchers with controlled access to health data. Typically built on the principles of cloud computing, the TRE stores de-identified data (A), associated with data management and curation software (B) and supported by dedicated computing and data storage facilities (C). The entire system is managed within a legal framework (D) with appropriate data security measures in place (E). The TRE will typically have secured certifications such as ISO 27001 to promote trust (F). Together, these features provide a tried and tested approach to streamlining and enhancing researcher access to healthcare data.

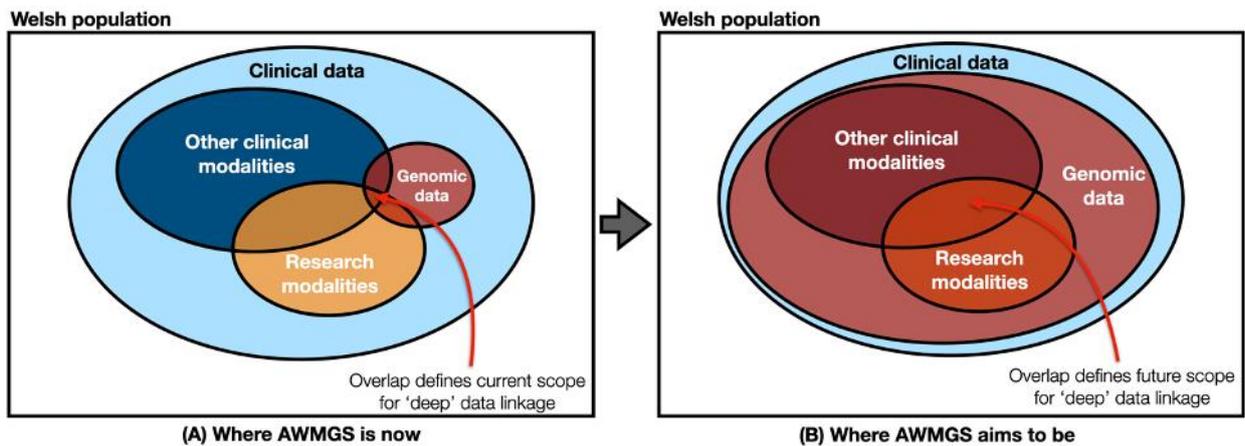
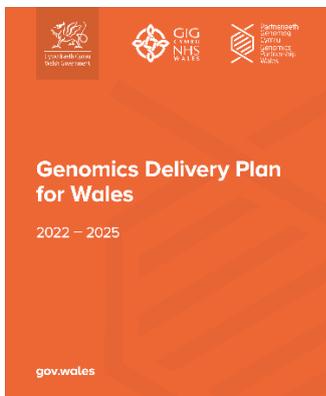


Figure 2: The value of genomic data for research will increase over time as more patients are sequenced. The difference between panel A (where we are now), and panel B (where we aim to be) represents the increase in genomic typing of patients planned by Genomics Partnership Wales and hence the potential expansion in added value achievable through data linkage. As the pool of sequenced patients increases, opportunities increase to link with other healthcare data such as medical imaging data and pathology reports. Equally genomic data can link with research data such as biobanking samples opening up new opportunities for research and innovation.

The Genomics Delivery Plan for Wales 2022-25 (<https://www.gov.wales/genomics-delivery-plan-2022-2025>) details how Genomics Partnership Wales will work with Welsh Government and other



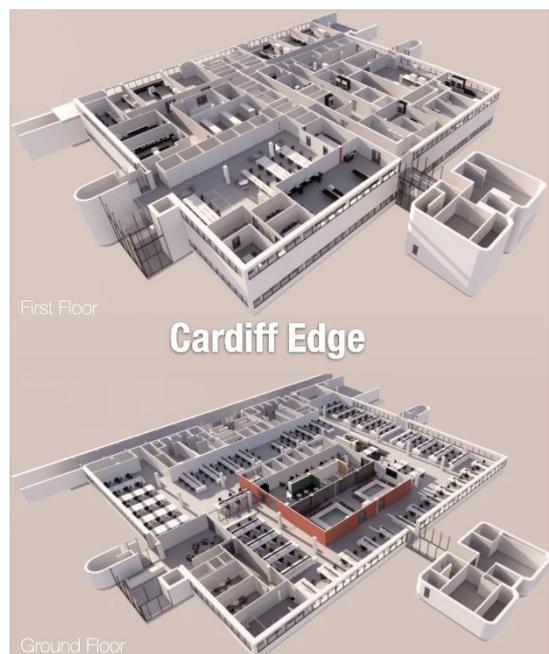
stakeholders, to harness advances in the understanding and application of genomics to transform public health strategy and delivery of care.

An important part of this delivery plan is the development of TRE services that will “facilitate researcher access to routine NHS genomic data through a consent system and development of a trusted research environment”

(Delivery Theme 3: Research and Innovation; Genome UK Pillar 3, Deliverable 4). To this end the delivery plan has several specific digital and

data deliverables that will enable Wales’ vision to be realised focusing around the digital infrastructure currently being designed for the new state-of-the-art genomics facility at Cardiff Edge Life Sciences Park, scheduled for the end of this year.

Wales Gene Park is a key partner in this development and through our own TRE programme (the Wales Cancer TRE Project, see Annual report 2022), we are actively working with our partners to deliver on thee digital and data deliverables and establish the Genomics Partnership Wales vision.



The Genomics Delivery plan has already committed Genomics Partnership Wales to developing links between AWMGS and the SAIL databank to ensure researcher access to anonymised processed genomic data. This is also a Wales Gene Park ambition and one of the aims of our Wales Cancer TRE Project so we are now seeing a coming together of ambition across several key organisations. Further TRE services will need to center around access to, and linkage of, raw genomic sequence data from routine genomic clinical tests. This poses more substantial technical, governance and security challenges that will now need to be resolved.

With this in mind, the partnership has developed a plan of work to develop the information governance, data security, and IT design needed to develop future shared digital services that will provide TRE services. This work is based around an initial investment in shared data storage hardware at Cardiff Edge, that we hope can get started later this year.

	Planned work	Year 1 2023-2024		Year 2 2024-2025		Year 3 2025-2026	
		H1 2023	H2 2024	H1 2024	H2 2025	H1 2025	H2 2026
		Technical implementation	Establish new data storage				
	Integrate partner systems						
Governance	Information Governance						
	Cybersecurity design						
	Accreditation and certification						
Technical design	Review of current setup						
	Craft strategic vision						
	Design of new system						
	Prepare next phase						
Next phase	Start next phase						

CASE STUDY 2

Wales SWAN UK Clinic

Developing evaluation tools for the Syndromes Without a Name (SWAN) Clinic Pilot

Genetic Alliance UK works collaboratively with Wales Gene Park, funding the Policy & Engagement Manager (Wales) post. The charity was awarded funding from the Lottery in 2020 to support the development of a SWAN network in Wales – SWAN UK Cymru. The SWAN network supports families affected by undiagnosed genetic conditions through peer-to-peer support, information sessions, outreach and health professional education.

We know life experiences can be very difficult for patient's living with a rare disease and, in particular, where they have not yet got a diagnosis. The patient journey crosses every part of the health service, which makes coordination of care difficult and includes a significant amount of help from social care and education, particularly for children.

In 2022, as a member of the Rare Disease Implementation Group with oversight for delivery of the Welsh Rare Disease Action Plan, Genetic Alliance UK supported a bid to Welsh Government. The bid aimed to establish a SWAN clinic pilot, for 2 years to develop an evidence base for commissioning an all-Wales SWAN service. The clinic sees paediatrics and adults, both virtually and in person and takes a multidisciplinary approach. Care coordinators were recruited to support patients and their families and coordinate care.

In addition to bringing together professionals working as part of the SWAN clinic, Genetic Alliance UK facilitated for the SWAN UK Cymru Sounding Board to help develop measures to evaluate the SWAN clinic, providing feedback on a suite of Patient Reported Outcome Measures (PROMs) and Patient Reported Experience Measures (PREMs) to be carried out with patients utilising the clinic. These measures will be key in learning about whether the clinic makes a difference to their life and if it does, how could this be improved?

The clinic links health, social care, education and research. Being involved in research is important for rare diseases patients. Wales Gene Park colleagues are working as part of the SWAN clinic, at the forefront of research. This will form part of the assessment of the clinic - finding new genetic variants, new conditions and writing up in scientific literature. Research will also be part of the outcomes developed.



It's about navigating the system; what is available to patients. Organisations such as Genetic Alliance, are a great help, involved in research and the knowledge of specialist conditions, but also organisations that can help patients in the social and education setting are key. *Dr Graham Shortland, SWAN Clinic Lead*

CASE STUDY 3

Genomics After Dark at Techniquest

Background

With genetics/genomics ever more vital in mainstream medicine and healthcare, public engagement and understanding in this area is vital, as reflected in Welsh Government's Genomics for Precision Medicine Strategy (launched 2017) and subsequent Genomics Delivery Plan for Wales 2022 – 2025.

As part of an ongoing programme of communication and engagement with the public around this subject, the inaugural Genomics After Dark outreach event was organised and delivered by Wales Gene Park's Education and Engagement Team in conjunction with Genomics Partnership Wales.

Held at the Techniquest Science Discovery Centre in Cardiff, the purpose of the event was to communicate with the public about genetics/genomics and showcase some of the work being carried out in this sector in Wales. It was aimed at an adult audience (particularly the hard-to-reach 18 to 50 age demographic) and free to attend.



Content

The event comprised a range of engaging, hands-on activities which included:

- Laboratory-based workshops, where attendees could learn about DNA and carry out DNA extractions
- Public talks focusing on ethics in clinical genetics/genomics, inviting audiences to consider a number of ethical questions and scenarios
- Virtual Reality games created by WGP and Genomics Partnership Wales
- Careers videos highlighting job roles
- A wide range of interactive stands from various organisations across sectors such as healthcare, research, public health, higher education and the arts, including: Wales Gene Park (DNA bracelets, DNA Snakes & Ladders, chromosome matching); Genomics Partnership Wales (genomics terminology game); Public Health Wales Pathogen Genomics Unit (interactive DNA modelling, bioinformatics); Inherited Tumour Syndrome Research Group - Swansea and Cardiff Universities (jelly 'mini-gut' organoids in personalised medicine); Cardiff University's Unique You Project (cancer myths/facts quiz and genomics in personalised medicine game); National Centre for Mental Health (Play Your Genetics Cards Right); Cardiff Metropolitan University (genetics of taste); Wales Cancer Research Centre (WCRC) and cancer research groups from Cardiff University (progression of cancer therapy, virotherapies); BRAIN Unit (Brain Games); Same But Different Rare Project (photography exhibition); All Wales Medical Genomics Service (genetics games and careers)



In addition, attendees could experience Techniquest's own hands-on scientific exhibits including planetarium tours.

Feedback and Future Plans

Genomics After Dark was attended by over 300 members of the public. Attendees were given the opportunity to give quantitative and qualitative feedback via evaluation forms and a feedback wall. Comments were also collected from exhibitors and social media interactions. The response was overwhelmingly positive (a full summary can be found in the event report), reflecting excellent engagement from visitors overall, with each aspect of the event being extremely well-received.

Following the success of Genomics After Dark, WGP has been asked by Genomics Partnership Wales to deliver repeat events in both north and south Wales during the coming year. Other outcomes have included a request for WGP's Education and Engagement Team to organise the Association of Genetic Nurses and Counsellors Annual Conference in Cardiff in 2024, several talks (for health professionals and public groups), and new members of WGP's Public Genetics Network and Rare Disease Patient Network.

I made DNA – AMAZING! Thank you.

Thank you for an awesome evening!

Great organisation and support for exhibitors.

Genomics is fun!

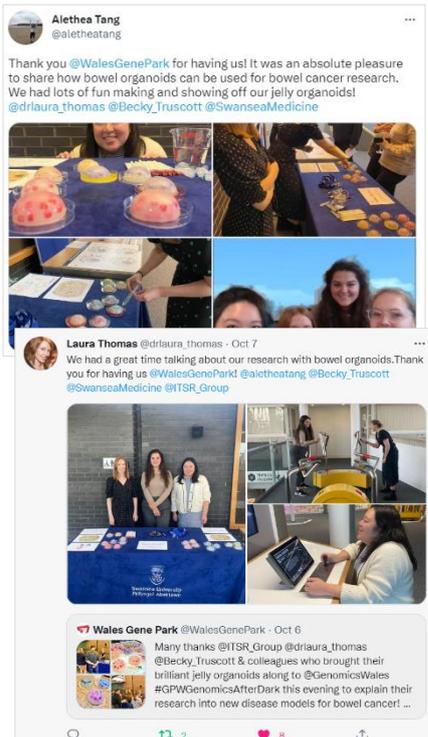


I love science!

Genomics is fascinating!

Very useful chat about career options.

Excellent event – keep them coming!



CASE STUDY 4

All Wales Wastewater-Based Antimicrobial Resistance Surveillance



Gordon Webster, Laura Rushton, Peter Kille, William Perry
& Andrew J. Weightman
School of Biosciences, Cardiff University

Wales Gene Park is facilitating sequencing metagenomic samples from across Wales to address the emerging AMR threat.

It is well established that the rise of antimicrobial resistance (AMR) represents one of the biggest threats to public health and healthcare provision of in Wales.¹ The issue of AMR has arisen due to the systematic misuse and overuse of antimicrobial drugs in human medicine and food production around the world, including Wales.² AMR is a drain on the Welsh economy with economic losses due to reduced productivity caused by sickness (of both human beings and animals) and higher costs of treatment.¹⁻⁴ This has led to the creation of the Welsh Antimicrobial Resistance Programme (WARP) with strategic aims and the formulation of a 5-year implementation plan to better understand the extent of the problem and help contain the increase of AMR.³ The approach is consistent with global efforts to reduce the impact and spread of AMR.⁵ The five key strategic objectives of the Wales 5-year AMR plan are (i) to improve awareness and understanding of antimicrobial resistance; (ii) to strengthen knowledge through surveillance and research; (iii) to reduce the incidence of infection; (iv) to optimize the use of antimicrobial agents; and (v) to ensure sustainable investment in countering antimicrobial resistance. In this pilot project we address specifically objectives (i) and (ii).

The establishment of the Welsh Government-sponsored national wastewater monitoring network during the COVID-19 pandemic has provided the infrastructure to undertake routine surveillance for a wide range of public health indicators in wastewater. To date, the samples collected from the network have been used to evaluate the amount of SARS-CoV-2 and other faecal indicator viruses in wastewater at 47 sites across Wales. This captures ca. 90% of the Welsh population. The benefits of the wastewater-based approach are that it provides an unbiased estimate of the amount of COVID-19 circulating in the population. After a successful pilot phase indicating that the approach can be readily applied to the surveillance of antimicrobial resistance genes (ARGs) circulating in the population, consistent with many other studies undertaken worldwide⁶⁻⁹, we are now undertaking the monitoring at a national level to support and inform government policy. In addition to the WasteWater treatment sites we have extended our surveillance to incorporate 10 hospital sites from across Wales. Currently, we collect and analyse samples of influent water (raw untreated sewage) at each wastewater treatment plant as our initial focus is on public health. However, at some sites pilot analysis is being performed to study both influent and effluent water to gain a better understanding of the efficiency of wastewater treatment across the national network as well as providing key information on the wider environmental impacts under the One Health agenda.

DNA extracted from the samples is prepared for Shotgun metagenomic sequencing undertaken through the NovaSeq Access through the Wales Gene Park. We have sequences samples monthly starting December 2022 with ~50 samples rising to >78 in May 2023. Bioinformatic analysis of the

data, performed at Cardiff School of Biosciences, allows us to capture a wider array of genes as well as identify microbial carriers of the genes. We are able to evaluate the abundance of clinically relevant taxa (e.g. *Salmonella* spp., *Acinetobacter* spp, *Escherichia-Shigella* spp., *Pseudomonas* spp., *Staphylococcus* spp. and *Vibrio* spp.). We are also able to identify the core plasmidome, virulome and mobilome of each sample. Currently, we are engaging stakeholders to design a dashboard that will allow relevant organisation to access and interrogate the data.

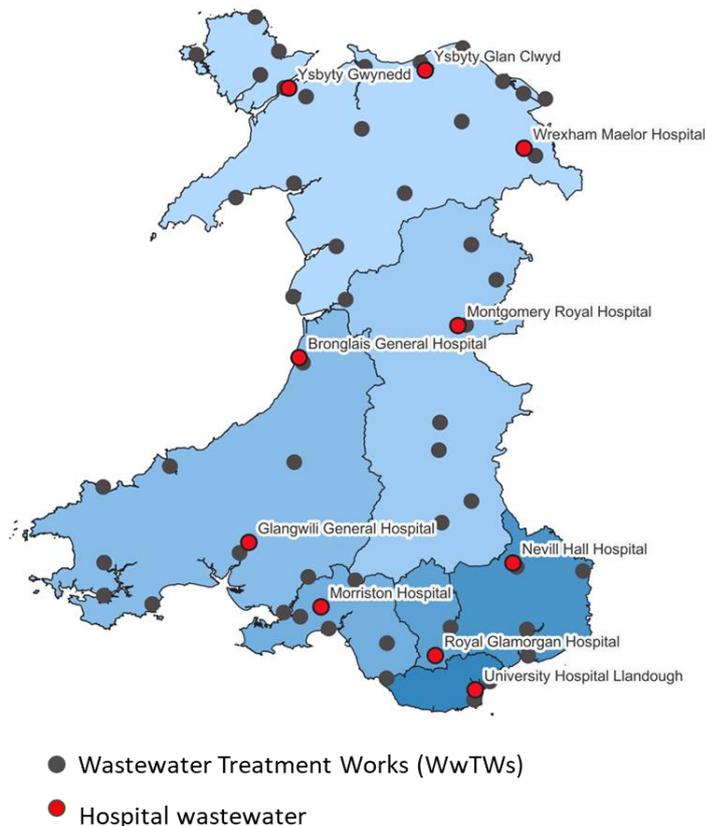


Figure 1: WasteWater and Hospital Sampling Sites

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LOOKING FORWARD

Supporting genomic research in Wales into the future

Wales Gene Park continues to build on our commitments to support delivery of Welsh Government's strategies, including the Genomics Delivery Plan for Wales 2022-25, and the Implementation Plan for Rare Diseases.

We will provide the technologies required by Welsh researchers:

- Partnership working to ensure the provision of cutting edge genomic and bioinformatic analysis techniques to support the needs of health researchers in Wales, including research undertaken in partnership with the NHS and with the commercial sector. These techniques will include long-read DNA and RNA sequencing, methylomic analysis and spatial transcriptomics.
- Human iPSC derivation and genome editing to support researchers needs through the provision of appropriate pre-clinical models of disease to understand the causes of disease and to develop and test potential treatments.
- Working with experts in e-health research at the SAIL databank at Swansea University to establish methods for integration of NHS genomic data with clinical and other e-health related data for research, innovation and service development.
- Consult with patients and the public to develop an open, transparent and publicly agreed approach to the sharing of genomic data for service development and research.
- Delivery of an extensive portfolio of events and activities to promote health professional and public education and engagement, and to facilitate the informed involvement and participation of patients and public in setting research priorities in health and social care and in research planning, funding, dissemination and translation.
- Service and policy development work, working with Genomics Partnership Wales, the new Welsh Parliament Cross Party Group for rare, genetic and undiagnosed conditions and other partners to influence and develop policy areas which improve the lives of patients and their families.

A time of change for WGP

Wales Gene Park has evolved since its initial founding in 2003 developing a strong reputation for its Education and Engagement activities, and as a major provider of high throughput sequencing and genomic analysis in Wales. In the next 12 months, Wales Gene Park will move along with the NHS All Wales Medical Genomics Service and the Public Health Pathogen Genomics Unit to form a prestigious new genomics hub based at the Cardiff Edge Business Park. Wales Gene Park will maximise the benefits of Welsh Government's investment in the new genomics hub with a focus on research and innovation. A key task over the next 2 years will be to work with stakeholders and funders to define our vision for Wales Gene Park going into the second half of the decade.

ACKNOWLEDGEMENTS

Wales Gene Park could not operate without the continued support from many others. Notably, many senior academics at Cardiff University, our colleagues with NGS expertise, the university management teams, and NHS staff, all of whom give their time, energy, and enthusiasm to support Wales Gene Park. We thank them all for their invaluable help.

We further thank all members of the Strategic Advisory Group chaired by Prof Colin Dayan, and our patient representatives, Mr Alan Thomas and Mr Nathan Davies, for their continued and excellent support and advice.

CONCLUSION

Genetics and Genomics are becoming ever more important for health research and health care. Wales Gene Park continues to work with Welsh Government, Higher Education Institutions, the NHS and industrial partners to make advances and to develop and deliver better quality health and care services in Wales.

Our commitment to support the Welsh Government's Genomics Delivery Plan for Wales, and the Implementation Plan for Rare Disease, will continue to inform our priorities and working practices. We are proud of the important role laid out for Wales Gene Park in the delivery of these strategies.

The provision of competitively priced, bespoke NGS and bioinformatics for researchers across Wales, is being further developed to enable long-read DNA sequencing and spatial transcriptomics.

The Education and Engagement team have delivered a large programme of events and activities to support the Genomics Delivery Plan for Wales. Current audiences include health care professionals, patients and families, schools and colleges, and the public. We also prioritise involvement of the public and patients, using our networks to ensure their views are communicated clearly and effectively to Welsh Government.

Wales Gene Park is an integral part of Genomics Partnership Wales and we will soon be moving to co-locate with the NHS All Wales Medical Genomics Service and the Pathogen Genomics Unit of Public Health Wales. This move will place Wales Gene Park at the centre of an exciting new Genomic Centre, well-placed to supporting researchers in accessing samples and data, and translating cutting-edge genomic techniques into new diagnostic tests.

GLOSSARY

A

AMR Antimicrobial Resistance

ARGs antimicrobial resistance genes

AWMGS All Wales Medical Genomics Services

C

CARIS Congenital Anomaly and Rare Disease Information Service

COVID-19 Coronavirus Disease

CPD Continuing Professional Development

CRISPR Clustered Regularly Interspersed Short Palindromic Repeats

D

DNA Deoxyribonucleic Acid

E

eFAD Early-onset Familial Alzheimer's Disease

ES Embryonic Stem

F

FAP Familial Adenomatous Polyposis

G

GPW Genomics Partnership Wales

H

HCM Hypertrophic Cardiomyopathy

I

ILAE International League Against Epilepsy

iPSCs Induced Pluripotent Stem Cells

ITSR Inherited Tumour Syndrome Research

M

MAP MUTYH-Associated Polyposis

MRC Medical Research Council

N

NGS Next Generation Sequencing

NHS National Health Service

P

PCD Primary Ciliary Dyskinesia

PEG Patient Empowerment Group

PenGU Public Health Wales Pathogens Genomics Unit

PPI Patient and Public Involvement

PROM Patient Reported Outcome Measures

PREM Patient Reported Experience Measures

R

RDIG Rare Disease Implementation Group

RNA Ribonucleic Acid

S

SAG Strategic Advisory Group

SAIL Secure Anonymised Information Linkage

SCE Specialty Certificate Examinations

SeRP Secure Research Platform

STEM Science Technology Engineering & Maths

SWAN Syndromes Without a Name

T

TRE Trusted Research Environment

TSC Tuberous Sclerosis Complex

U

UHB University Health Board

UK United Kingdom

UKRI United Kingdom Research and Innovation

USA United States of America

V

VCF Variant Call Format

W

WCRC Wales Cancer Research Centre

WGP Wales Gene Park

WARP Welsh Antimicrobial Resistance Programme