

ANNUAL REPORT 2020-2021

WALES GENE PARK



Wales | PaRC
GENE | GENETEG
PARK | CYMRU



Ymchwil Iechyd
a Gofal Cymru
Health and Care
Research Wales



Ariennir gan
Lywodraeth Cymru
Funded by
Welsh Government

2020 - 2021
Annual report



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“Harnessing Genetics and Genomics to advance Research, Healthcare, Education and Innovation”



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Welcome

Annual Report 2021



Dr Karen Reed

Co-Director and Operations
Manager, Wales Gene Park

Wales Gene Park is an infrastructure support group funded by Welsh Government through Health and Care Research Wales.

Welcome to the Wales Gene Park annual report which summarises Wales Gene Park activities during the period 1st April 2020-2021.

The 12 months of activity covered in this annual review has, as it has for many organisations, had its challenges. New ways of working are now embedded in our practices to accommodate revised working practices enforced by the COVID-19 pandemic. For a short time, our lab teams were unable to access the laboratories, but we are pleased to say these are now operating as normal. Despite the challenges faced, we are proud that Wales Gene Park has continued to work to provide collaborative access to genomic technologies for researchers in Wales. All Education and Engagement activities have moved online to use virtual platforms to continue to provide innovative initiatives to educate and engage health professionals and the public in Wales and beyond.

As an integral partner within Genomics Partnership Wales (GPW), we have been committed to developing the plans for a new state of the art facility to enable the three GPW partner organisations to co-locate to deliver an integrated genomics service: All Wales Medical Genomics Service, Pathogen Genomics Unit and Wales Gene Park. Together, we strive to deliver activities that aid the implementation of the Welsh Government's Genomics for Precision Medicine Strategy to benefit the people of Wales.

Foreword

Harnessing Genetics and Genomics to Advance Research, Healthcare, Education and Innovation



Dr Andrew Fry
Director, Wales Gene Park

This annual report sets out the value and impact of the extensive portfolio of work undertaken by the Wales Gene Park.

Genomics is central to contemporary biomedical research and has played key roles during the COVID-19 pandemic. Wales Gene Park education and engagement activities have included numerous public events exploring the role of human and viral genomics in the pandemic, and explaining how genomics contributed to the rapid development of COVID-19 vaccines. In addition, Wales Gene Park bioinformaticians have supported researchers analysing viral genomes to develop analysis tools to track different variants of the virus.

The past 12 months have been challenging for everyone including the Wales Gene Park team, however we have continued to provide support to researchers working on cancer, mental health, infectious disease, rare disease, and others. This year we have generated sequencing data for 29 genomic projects, provided bioinformatics support for 33 projects and supported external grants bringing in over £5M to Wales.

Despite the challenges this report demonstrates the breadth, quality and quantity of work undertaken by the Wales Gene Park during the year 2020-2021. We have all found new ways of working over the last year and I have been impressed by how the Wales Gene Park team have analysed genomic data remotely, engaged and educated virtually, and performed socially distanced laboratory work. I hope we retain the best aspects of this experience going forward.

A word from our Patient & Public Representatives

A new funding period has coincided with a new patient representative as we welcome Nathan Davies as a new patient representative within Wales Gene Park. We wish to formally thank Perry James for all he has undertaken to help Wales Gene Park for the previous 5+ years. Our patient representatives offer valuable insight to shape our strategic direction. Nathan will join Alan Thomas for this role.

Introducing Nathan Davies, a new patient representative within Wales Gene Park.

“I have been involved in Patient and Public Involvement (PPI) activity in health and care research for about 10 years. For example, I have reviewed research grant applications for the National Institute for Health Research (NIHR) and have been a member of various groups, such as the Health and Care Research Wales Scientific Board for Health Research Grants. I have reviewed curriculum Modules for the Health Scientist Training Programme, administered by the National School of Healthcare Science (NSHS). I have also reviewed public-facing materials, such as information packs and consent forms for randomised clinical trials.

I enjoyed a diverse career in education before working in the field of sight loss (third sector organisations) for a number of years. I am also a qualified project manager and have managed and delivered health, education, food and drink and heritage projects.

I enjoy spending time with my family and going for walks along one of our many local beaches.”

Wales Gene Park

Introduction



Introduction

Wales Gene Park is an infrastructure support group 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 seamless translation of advances in genetics and genomics to improve NHS services and commercialization.
- Ensuring the informed involvement of patients, public and professionals in the development of genomic medicine in Wales.

Wales Gene Park

Strategic Objectives

Expertise and infrastructure

To develop and provide expertise and infrastructure that will help Wales compete at the forefront of genetic and genomic research in its areas of priority and strength, in particular by supporting Health and Care Research Wales funded research activities and programmes.

Support Welsh researchers

To support Welsh researchers through the provision of conventional and novel applications of Next Generation Sequencing, transgenic and genome editing technologies, focusing on areas of research and clinical expertise and patient needs in Wales.

Drive the development of Genomic Medicine in Wales

To drive the development of genomic medicine in Wales, focusing particularly on translational genetic research in common and rare disorders in which Wales has research strength.

Leadership

To provide leadership in rare genetic disease research by helping set priorities, engaging patients and families, promoting research collaboration using existing Welsh Government, NHS and Higher Education Institution mechanisms and by creating stronger links with industry.

Help Wales develop capacity in genomics

To help Wales develop capacity to undertake genomic analysis on a significant scale and support the linkage of genomic information to clinical data and other relevant data sets.

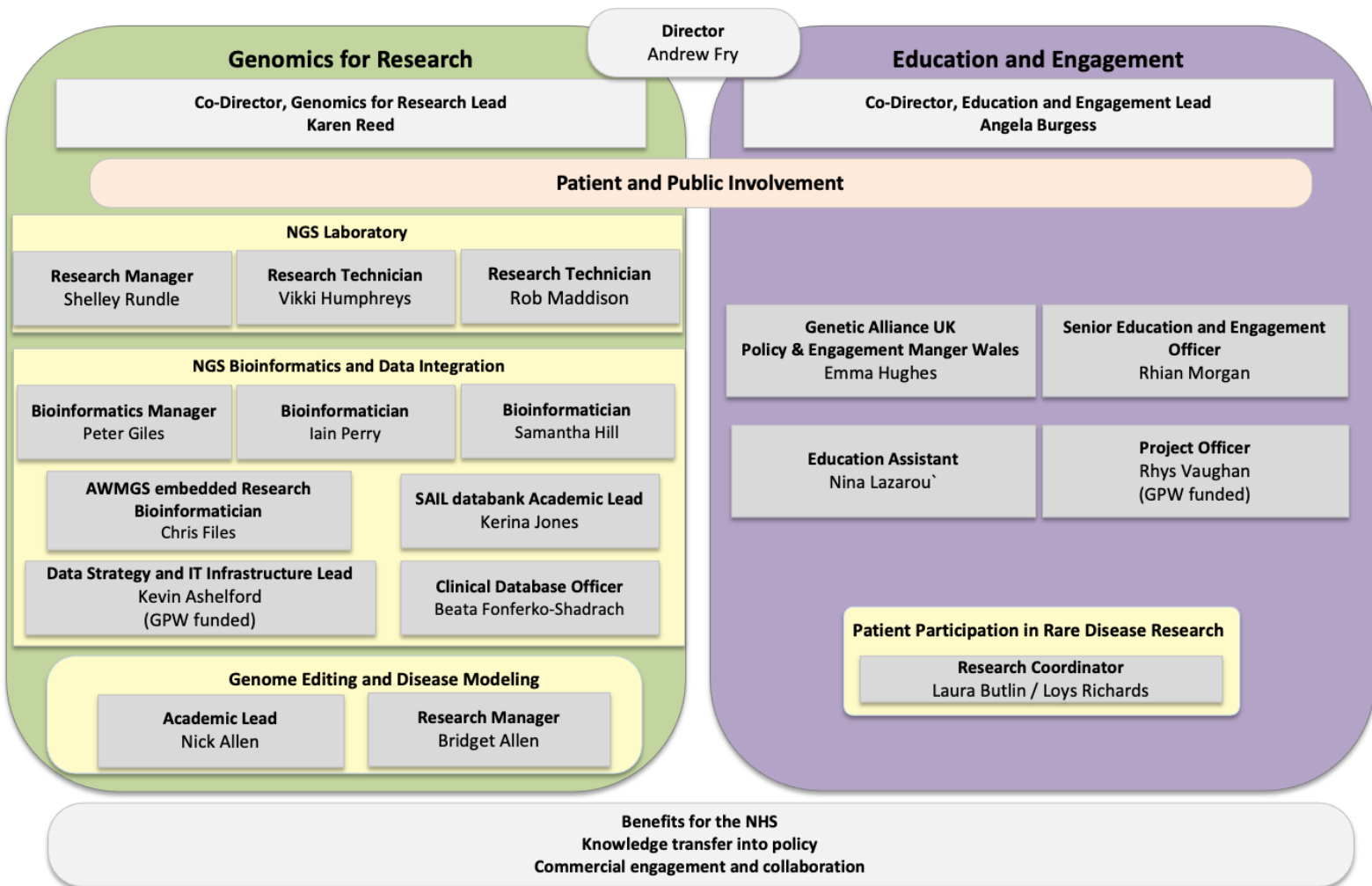
Awareness and understanding

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 Wales Gene Park Structure

The Wales Gene Park annual budget employs 20 staff (13.9 full time equivalent), who are supported by senior academics, experts from the NHS in Wales, managers and administrative staff from the host institution, Cardiff University.

Wales Gene Park Structure April 2020-April 2021.



Who's Who and Key Partners 2020-2021

Academic Partners

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

NHS Partners

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

Commercial Partners

- Agilent Technologies
- Cellesce Ltd.®
- GW Pharmaceuticals plc
- Illumina
- New England BioLabs, (UK) Ltd
- PTC Therapeutics

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
- Techniquist
- Tenovus Cancer Care
- The Wellcome Trust
- Tuberous Sclerosis Alliance (USA)
- Tuberous Sclerosis Association (UK)

Strategic Advisory Group

- Chair: Professor Colin Dayan, School of Medicine, Cardiff University
- Dr Rob Orford, CSO Health, Welsh Government
- Dr Ramsey McFarlane, Bangor University
- Professor Malcolm Mason OBE, College of Biomedical and Life Sciences, Cardiff University
- Jayne Spink, Chief Executive, Genetic Alliance UK (Vacated role mid reporting period)
- Dr Mick Hunter, Entrepreneur in Residence Evotec, COO Viatem Ltd, and CEO Orca Pharmaceuticals Ltd
- Dr Clive Morgan, Cardiff and Vale University Health Board
- Michaela John, Genomics Partnership Wales.
- Sian Morgan, Head of Laboratory at the NHS All Wales Medical Genomics Service Laboratory
- Joanne Ferris, Operations Manager The Association of the British Pharmaceutical Industry
- Mark Bale, Head of Science Partnerships, Genomics England

Patient Representative

- Mr Alan Thomas

Executive Management Team 2020-2021

- Dr Andrew Fry, Director
- Dr Karen Reed, Codirector, Operations Manager, Genomics for Research Lead
- Mrs Angela Burgess, Codirector. 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, Genetic Alliance Policy and Engagement Manager, PPI Lead
- Professor Andy Tee, Commercial Lead

Lay Summary

Wales Gene Park is an infrastructure support group funded by Welsh Government through Health and Care Research Wales. During the 2020-2021 reporting period we supported research via two work packages.

Work Package 1

Geonomics for Research

Sequencing and analysing genomic information in collaboration with researchers.

Making and using pre-clinical models of disease through genome editing and related methods.

Work Package 2

Education and Engagement

Enhancing awareness and understanding of health-related genetic and genomic research for the public and health professionals.

Influencing Welsh Government Policy development.

Summary of activities

Developing models of disease

Before new treatments can be trialled in the clinic, evidence from pre-clinical work is needed. At Wales Gene Park we help researchers generate new genetic models of disease to undertake this pre-clinical work. We have supported 7 projects for 7 research groups in the areas of neuroscience, epigenetics, optometry, and cancer.

Clinical Research

Access to patient samples and clinical data is essential for clinical research. Wales Gene Park manages the study governance, patient recruitment and organising of study data for 4 open/active UK Clinical Research Network (UKCRN)/Health and Care Research Wales Clinical Research Portfolio projects in rare diseases. Through obtaining 22 new patient consents and banking 88 samples from patients with cancer or rare diseases, we have supported four laboratory-based research projects that aim to improve future NHS diagnostics.

Sequencing provision

Next Generation Sequencing (NGS) is a powerful, cost-effective, time-efficient technique used 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 supported 29 research projects, 59% of projects were for cancer biology and/or rare disease-related research, the rest were for other common diseases including arthritis, kidney disease, immunological conditions, neurodegeneration and Alzheimer's projects.

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 33 research projects this year and provided access to computing capacity to a further 28 researchers and postgraduate students. Wales Gene Park is now custodian to over 384 terabytes of sequencing data held on behalf of researchers across Wales.

Education and Engagement

The Wales Gene Park Education and Engagement team run many events to raise awareness and understanding of the advances of genomics within our health care system. This year, through 59 events organised by Wales Gene Park (a further 22 were cancelled due to COVID-19), we have reached:

- 999 Health Professionals: 22 events
- 146 Schools/Colleges students/teachers: 2 events
- 2275 Public/Patients: 39 events

Supporting Policy and Working with patients with rare disease

This year has been particularly challenging for the rare disease community, many of whom have been shielding during the COVID-19 pandemic. Our work has centred on ensuring that patients and families continue to feel engaged and supported and their voice is heard by decision makers. We have engaged and involved people affected by rare conditions in our policy work and research activities through virtual Genomics Cafés and the Cross Party Group on Rare, Genetic & Undiagnosed Conditions. We have recruited a new Patient and Public Representative to the Strategic Advisory Group (SAG) this year. We have also offered 18 opportunities to our rare disease patient network to be involved in research and 146 individuals have taken up these opportunities.

Core Metrics

Reporting period: **2020/2021**



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



Direct
funding
awarded
£698K

Jobs created
through direct
funding



Grants won during reporting period

Grants won	Led by group	Group collaborating
Number	6	11
Value	£288,104*	£4,894,119
Funding to Wales	£288,104*	£4,786,264
Funding to group	£120,283*	£49,039
Additional jobs created for Wales	2	14
Additional jobs created for group	2	



Number of publications



Number of public
engagement events



Number of public
involvement opportunities

*Note, these values include £115,000 awarded by Welsh Government to enable the implementation of the Genomics for Precision Medicine Strategy in Wales



Supporting research across Wales and beyond

Work Package 1: Genomics for Research

Purpose

Wales Gene Park supports and promotes the application of genomic technologies via a range of activities delivered through a Next Generation Sequencing (NGS) laboratory, Bioinformatics provision, 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 to flexible NGS for research, together with expert bioinformatics support underpinned by high-performance computer infrastructure dedicated 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.

Appropriate pre-clinical models of disease are essential for biomedical research. While these models can take several forms, at Wales Gene Park we have expertise using patient-derived cell lines including human induced pluripotent stem cells (human iPSCs) for research. iPSC technology puts patients at the heart of functional genomics research because these cells can be generated from a patient's or control's own cells such as skin cells. Genome editing within iPSCs enables researchers to investigate the causes of disease and provides a platform for testing potential therapies.

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 SAIL databank (Swansea)
- Contribution to healthcare professional education
- Derivation and Growing human iPSC cells
- Utilising CRISPR (Clustered Regularly Interspersed Short Palindromic Repeats) genome editing technology to generate new models of disease for research
- Embryonic Stem (ES) cell derivation from genetically modified mouse models
- Consultation for researchers requiring support in ES cell and genome editing techniques

Progress

Wales Gene Park Preclinical modeling of disease

While COVID-19 operating procedures resulted in the closure of the genome editing lab for a short while, the small team have continued to enable research using human induced pluripotent stem cell (human iPSCs) for pre-clinical modeling of disease. 7 research groups have been supported in the reporting period, either through experimental design for grant applications, the provision of training, or establishment of novel lines of genetically modified human iPSC cells.

Prof Yves Barde, FRS, Cardiff University commented “WGP’s help proved invaluable in the context of a key project... their generosity with reagents meant that the process ran quick and smoothly.”

Wales Gene Park Genomics Facility

Despite a temporary closure of lab activities, the Genomics Facility has provided NGS data for 18 research projects, and processed samples for a further 9 projects and access to specialist equipment for 2 others, supporting 29 different research groups overall. The majority of projects, 59%, were for cancer biology and/or rare disease related research. Research for a range of other common diseases including arthritis, kidney disease, immunological conditions, neurodegeneration and Alzheimer’s projects accounted for the remainder. During the year, the genomics facility laboratory has received £44,535 to cover the costs required to fund the work undertaken.

Additionally, support and quotes for work were provided for 46 projects and 27 grant applications.

Wales Gene Park Bioinformatics

The Wales Gene Park Genomics Bioinformatics team provided analysis and other support for 33 projects during the 2020-2021 reporting period. More in-depth bespoke analysis was provided for 24 projects (73%), and 9 projects (27%) involved quality control analysis of sequencing data before the data was sent to researchers to perform their own analyses.

40% of analysed projects were for cancer research, 27% involved rare disease research, 15% concerned infection and immunity with the final 18% comprising a mix of neurological, arthritis, wound healing and musculoskeletal disorders. 30 out of the 33 projects have involved analysis on data generated in-house by the Wales Gene Park Genomics Facility Laboratory.

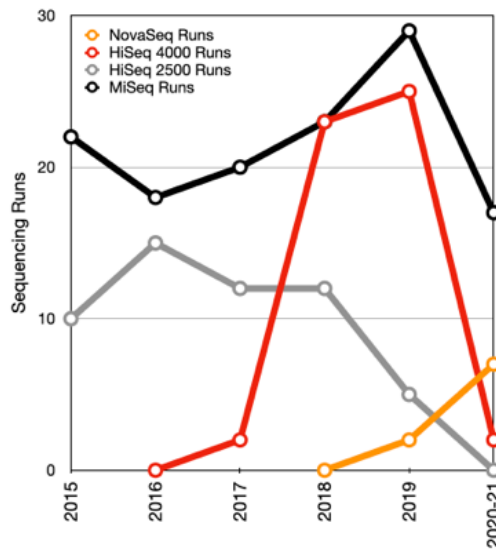
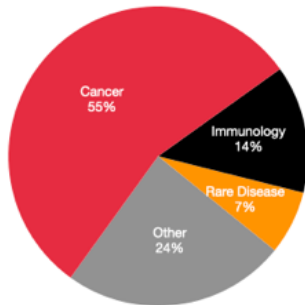
Project highlights and novel analysis methods employed by the bioinformatics team over the last year have included:

- Pilot work exploring immune signatures in publicly available single cell RNAseq datasets from COVID-19 patients.
- Forging a new collaboration with researchers in the Bristol Veterinary School using machine learning methods to identify potential immune signatures in human osteosarcoma patients.
- Working with researchers in the European Cancer Stem Cell Research Institute to characterise sequence variants in patient derived xenografts models of breast cancer.
- The development of commercial involvement in a previous project looking at machine learning techniques to improve detection in prostate cancer. This project combines the expertise and knowledge between Wales Gene Park, Atos, Swansea University and Supercomputing Wales.

Flexible NGS for medical genetics and genomics research

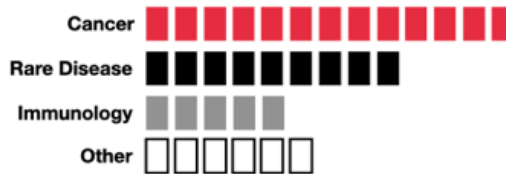
Sequencing

29
Projects



Bioinformatics

33
Projects



Supercomputing Infrastructure Users

28



360Tb WGP NGS data



384Tb WGP storage used by others

Wales Gene Park Dedicated compute and storage

IT underpins the use of genomic technologies so we are always looking ahead to ensure future genomic research is properly supported. This is why we have partnered with Wales Cancer Research Centre (WCRC), Wales Cancer Bank (WCB), Cardiff University School of Medicine, and the All Wales Medical Genomics Service (AWMGS) among others, to build a new compute and data storage facility that is dedicated for cancer and rare genetic research. In 2020 we secured a Sêr Cymru Infrastructure Accelerator Award for £277,865 to do just that and, working with ARCCA, we are currently in the process of building this IT resource to be ready for use later this year.

This new resource will compliment and supercede the existing Wales Gene Park compute resources that are used for Genomics research, hosted by Supercomputing Wales. During the reporting period 18 researchers and 10 postgraduate students, have made use of this dedicated resource for genomics, bringing the total number of researchers and students who have been granted secure access to these resources to support their research to 219. Wales Gene Park is now custodian to over 360Tb terabytes of sequencing data held on behalf of researchers across Wales and has provided 384Tb terabytes of storage to other Welsh research groups for the storage of their genomics data.

The new Sêr Cymru IT resource is being designed with higher levels of data security than we've previously needed so that we can better support the integration of research data with routinely collected clinical data from our NHS partners. This will help us realise the ambition of Genomics Partnership Wales to improve data sharing for patient benefit.

As examples, as a direct result of the Sêr Cymru IT we are now working with WCB, WCRC and the AWMGS to sequence cancer tumour biopsy samples and link these data to histology images to create a combined data repository that will eventually support the development of new AI technology to better stratify cancer patients for more personalised treatments. We are also supporting a project to help develop an improved risk stratification model for identifying individuals at high-risk of colorectal cancer, and another with the Marie Curie Palliative Care Research Centre to help develop their SMART-MDT strategy that should eventually lead to a decision support system to support care of oesophago-gastric cancer patients.

Enhancing understanding of genetics and genomics throughout Wales



Work Package 2: Education and 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 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), with increasing membership numbers in each group
- 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

The Public

A range of activities and events were held for public and patient groups. These included: Lay talks on DNA and genetics/genomics-related topics, delivered to **community-based groups** throughout Wales such as The University of the Third Age, The Rotary Club, Women's Institute groups, Probus Clubs, and library groups throughout Wales; **The 5th Annual Genetics & Genomics for the 3rd Generation (3G) public conference**, delivered in a virtual format for the first time; **a public lecture series** - aimed at engaging with a lay audience about genetics and genomics-related subjects – which comprised a virtual lecture on rare cancers, held in conjunction with the Wales Cancer Research Centre to coincide with Rare Cancers Day on October 1st; a talk on adopting technologies from sectors like genetics, biotechnology and medicine for the ongoing mission to Mars, held as part of Cardiff University School of Medicine's Science in Health Public Lecture Series; a virtual lecture entitled *V for Vaccination: from Cowpox to Covid* in collaboration with Integrated Wellbeing Networks Gwent (Aneurin Bevan University Health Board) which comprised a talk on the social history of vaccination and an interactive session on what it means now for COVID-19. Such was the strong public interest in the subject and high demand for places, this session was repeated reaching over 560 people.

"Thank you to all the speakers their enthusiasm for their work is what gives us hope for changing treatments and improving the lives of people effected with rare cancers"

Attendee, Rare Cancers talk



"Thank you so much for a most informative, enjoyable and uplifting evening talk. My first with you!"
Attendee, V for Vaccination talk

Public Genomics Cafés form part of the Genomics Partnership Wales engagement programme, that Wales Gene Park oversees. These are free and open to all; they include short, relaxed talks about genomics relating to health and medicine, and guest slots from health professionals, researchers and those affected by genetic and rare conditions, who share personal experiences.

In October 2020, a Young People's Genomics Café - primarily aimed at the 16 to 25 age group - was launched. Two events have been held to date, attracting over 225 attendees. For further details of the Genomics Cafés, please see the case study 5 (page 33).

Schools and Colleges

Although activities for Schools and Colleges were affected severely by the COVID-19 pandemic WGP continued to deliver on, and develop, several aspects of its programme including delivering the *Teachers' Genetics Network newsletters*, running a virtual 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*, developing a new *WGP Genomics Podcast* and a collaborative project with Illumine Theatre and Theatr Genedlaetho to produce a *Podcast drama*.

Health Professionals and Researchers

In autumn 2020, WGP launched its *Professionals' Genetic Network*, aimed at health professionals and those from research and academic communities with an interest in Genetics and Genomics. Members receive a biannual e-newsletter with relevant information and news from WGP.

Throughout the year, several events were organised for health professionals and researchers working in genetics and genomics. These included an *Artificial Intelligence (AI) and data integration workshop* exploring how data integration and AI can better exploit medical big data for research and healthcare; a *Familial Paranglioma Syndromes and their Clinical Management All-Wales Multidisciplinary Virtual Study day* open to those wanting to learn more about familial paraganglioma syndromes; a virtual *Rapid Next Generation Sequencing Diagnostic Workshop* which considered the future applications for rapid diagnoses and bioinformatic challenges; the *annual Clinical Genetics Revision Course* for doctors training and specialising in this area of medicine, hosted by the All-Wales Medical Genetics Service.

Four *Living with Genetic Conditions* sessions which comprise talks from a Genetic Counsellor and individuals affected by genetic conditions, delivered for various healthcare professional trainees at Welsh universities, as well as several other educational sessions for post-graduate students and researchers.

Several other educational sessions were also delivered to Cardiff University Division of Cancer Genetics post-graduate students and researchers.

The Education and Engagement Team also gave virtual poster presentations at the European Conference for Rare Diseases in May and at the annual Health and Care Research Conference in October. Presentations were '*Engaging and Involving the Rare Disease community in Wales through Genomics Cafés*', '*Co-production of the Welsh Rare Disease Research Gateway*' and '*Making a Difference: working with those affected by Rare Diseases in Wales*'.

Genomics Partnership Wales Genomics Roadshow talks offered health professionals opportunities to learn about advances in genomic medicine in Wales, including Welsh Government's Genomics for Precision Medicine strategy. They are delivered throughout Wales by experts from the All-Wales Medical Genomics Service. Despite being severely disrupted by the COVID-19 pandemic, virtual sessions were given to health professionals at Withybush and Nevill Hall Hospitals, and to year 1 and year 3 undergraduate midwifery students at Cardiff University.

Patients & Families

New project funded - SWAN UK Wales - July 2020

Funding of a new project - Syndromes Without a Name (SWAN UK) Wales/ Cymru. The Big Lottery funding of £162,821.00 will be provided over 3 years.

SWAN UK is the only dedicated support network for families with children who have undiagnosed genetic conditions. SWAN UK – CYMRU aims to:

- Identify and engage more Welsh families into the existing SWAN UK – Cymru peer support network so that they feel less isolated.
- Deliver bilingual information, support, activities and events specifically tailored to the needs identified by Welsh families.
- Network with local services and organisations to help educate local professionals on the needs and challenges faced by SWAN UK families in Wales and the vital importance of effective care coordination.
- Develop and produce a bilingual early intervention toolkit – Rare Resources Wales – which will provide a wide range of general information on genetic, rare and undiagnosed conditions as well as advice on how to access reliable information, care and support in Wales.

A National Lottery Community Fund news article announcing the funding can be accessed here:

<https://www.tnlcommunityfund.org.uk/news/press-releases/2020-08-06/communities-in-wales-awarded-more-than-15-million-national-lottery-funding-since-start-of-pandemic>



*“I was so glad to find SWAN UK! Suddenly we no longer felt alone. I couldn't believe how many other families were going through the same thing as us’
Tammy from Bargoed near Caerphilly.*

The annual *Rare Disease Patient Network meeting - December 2020*, was held virtually and was attended by 71 people affected by rare and genetic conditions. The meeting included the launch of Rare Experience 2020 - a report based on a research project conducted by Genetic Alliance UK with over 1000 responses from the rare disease community:

<https://rareexperience2020.geneticalliance.org.uk/wp-content/uploads/2020/12/Rare-Experience-2020-Report-.pdf>

Wales Gene Park has also worked with Genetic Alliance UK to **support the rare disease community throughout COVID-19**. In June 2020 weekly community check in meetings for member organisations to come together and share experiences from their communities. The check-ins have featured 4 talks from Welsh speakers including:

- Helen Iliff, an anaesthetist working for the Bevan Commission who developed a social distancing badge that is now widely available throughout Wales.
- An update from the All Wales Medical Genomics Service on the rollout of genomic medicine services.
- Health Technology Wales inviting patient organisations to get involved in supporting health technology appraisals.
- Pairing of MSc Genomic Counselling students from Cardiff University with patient organisations to develop research projects.

Genetic Alliance UK also launched a new, accessible COVID-19 Hub which provided up to date information for individuals, parents/ carers and organisations and covered topics such as health and wellbeing, education, and finances. It provides condition specific and country specific support:

<https://covid-19.geneticalliance.org.uk/wales/>

The **Annual Rare Disease Day Parliamentary Virtual Reception** brought together all four nations of the UK and had representation from parliamentarians, policy leads and patients and carers affected by rare and genetic conditions. The event focused on the publication of the new UK Framework for Rare Diseases and how each nation planned to implement its recommendations through their own Action Plans.

Chair of the Rare Disease Implementation Group, Dr Graham Shortland spoke about Wales' plans for implementing the Framework and two patient speakers highlighted their experiences of life with Turner's Syndrome.

Enabling Policy

Cross Party Group meetings

The Welsh Cross Party Group (CPG) for Rare, Genetic & Undiagnosed Conditions held its inaugural meeting in the Senedd in September 2019. Over the last 12 months, the group chaired by Angela Burns MS and administered by Genetic Alliance UK and Wales Gene Park, has moved to a virtual platform, and the experiences of those affected by rare and genetic conditions about the impact of the Covid-19 pandemic have been fed to Welsh Government through the third sector stakeholder forum.

The CPG produced a report launched at a parliamentary meeting in February 2021 highlighting Rare Disease Day. The report documents strengthening the patient voice in decision making processes and policy development, collaborating with the National Centre for Mental Health to support the dissemination of research to provide an evidence base for service developments in genomics, and improving research opportunities for those affected by rare and genetic conditions through linking researchers with NHS SWAN clinics to support research into undiagnosed genetic conditions following a negative result from genomic sequencing.

The full report is available here:

<https://geneticalliance.org.uk/gauk-news/news/wales-cpg-report/>

Other work involved input to a Medicines and Healthcare products Regulatory Agency Patient Group Consultation Event on Regulatory Flexibilities during COVID-19 and a consultation event held by the Department of Health and Social Care regarding feedback on the initial draft of the new UK Framework for Rare Diseases. Wales Gene Park also continues to input as a patient representative member to the Rare Disease Implementation Group (which has oversight for the Welsh Action Plan and Genomics Partnership Wales Programme Board. Throughout the last year, the patient experience during the pandemic was fed in via the Rare Disease Implementation Group lead and lead commissioner for the Welsh Health Specialised Services Committee. Other issues were also fed in via the Welsh Government Third Sector Stakeholder Forum.

Genomics Partnership Wales Patient & Public Sounding Board consultations

WGP worked with Genomics Partnership Wales to recruit the second cohort of 11 new members to the Patient & Public Sounding Board which provides patient and public advice and input to the work of the Genomics of Precision Medicine Strategy and its programme of work.

Five meetings of the Board were held in the reporting period 2020/21 and 2 in the reporting period for 2021. Topics included: Input to the design and development of a new estates building bringing together genomic testing, clinical facilities and genomic research in Wales and feedback on patient and public involvement in other programmes including pathology, imaging and advanced therapies.

Coproduced with members, this [video](#) has been created to showcase the impact members have made through being involved with the Board.

Involvement in Research

Wales Gene Park facilitated several opportunities for patient and public involvement in research over 2020 - 2021. Throughout the year, we offered opportunities to our rare disease patient network to become involved with shaping research studies, participate in consultations to support genomic research and to develop and guide workshops and studies that were being undertaken by research groups. In total, 146 individuals have been involved in genomic research during that period.

A new Patient and Public Involvement Representative was recruited to the Wales Gene Park Strategic Advisory Group and an induction session was held with both PPI Representatives who sit on the Group.

Linking those living
with genetic conditions
to advances in genetics
and genomics research

Achieving Impact

The Wales Gene Park mission is to support and promote genetic and genomic research across Wales to help create a sustainable, internationally competitive environment for genetic and genomic research, ultimately leading to improved health, wealth, and prosperity for the people of Wales.

Collaborative working across a range of healthcare specialties underpins Wales Gene Park activities. The following exemplars demonstrate the impact achieved from this way of working:

- Genomics for public health (Case Study 1). Translating the latest research techniques into applications that directly impact patient wellbeing is a core goal of Wales Gene Park. To realise this endeavor, we have embedded a bioinformatician with the All Wales Medical Genomics Service to test whether the latest genomic sequencing analysis techniques can improve the interpretation of patient data, while another of our bioinformaticians has been working with Public Health Wales to develop a rapid test to identify COVID-19 outbreaks using samples of wastewater.
- Recognising the growing need for appropriately trained bioinformaticians, Wales Gene Park supports a programme of specialist training to help plug this skills gap (Case Study 2).
- Working with commercial partners is an essential route to drive research discoveries through to novel therapeutics. Case Study 3 outlines the support Wales Gene Park provided Prof Simon Reed, helping him establish a new biotech start-up company.
- Through our close collaboration with the SAIL Databank we continue to explore ways of safely linking genomic data to other anonymised epidemiological information for population research benefit (Case Study 4).
- To achieve the greatest impact for patients with rare and/or genetic diseases and their families, Wales Gene Park provides a range of events to disseminate new research findings and promote the role of research around genomics and disease. The success of the Genomics café programme is highlighted in Case Study 5.

Case Study 1

Genomics for public health

Collaboration and adaptation underpin vital components of Wales Gene Park's activities. With our partners we strive to deliver a positive impact to Welsh public health, enabling the translating cutting-edge academic research into clinical settings.

Responding to a global pandemic

The emergency of the new global pandemic led to changes in focus for all. Within Wales Gene Park we have adapted our activity to support research around and understanding of coronavirus.

Data Analysis

Our bioinformatics team have been playing a leading role, in collaboration with Public Health Wales, to develop a Next Generation Sequencing analysis platform to detect and monitor COVID-19 variants from wastewater samples. This early-warning test is critical to monitor COVID-19 levels at a population level, allowing rapid containment procedures to be employed should the virus be detected.

The team have also been involved in developing optimised analysis methods to identify variants from samples collected as part of Cardiff University's screening service, adapting methods for both new tests and screening existing data that has been collected over the last year to explore the impact of variants of transmission within our staff and student populations.

Furthermore, the bioinformatics team worked with Prof. Girish Patel, Cardiff University, to undertake pilot work looking at the immune signatures of COVID-19 patients. This involved the mining of several public datasets which looked at the gene expression profiles from individual cells in patients with coronavirus. The analysis undertaken contributed to a grant application looking to targeting COVID-19 infection by reviving the antiviral cell response.

Education and Engagement

Our education and engagement team have run events with a strong COVID-19 component, thereby raising public awareness and understanding. Events with expert speakers highlighted the necessary adaptations and impact of COVID-19 for rare disease patients, offering both practical advice and creating a forum for the rare disease community to share experiences. The impact of the pandemic on healthcare delivery, the required adaptations and the establishment of the Dragon's Heart Field Hospital have been highlighted and events looking at aspects of immunology and the history of vaccination demonstrated how existing knowledge is being adapted to tackle coronavirus.



Working with the NHS

The Wales Gene Park Award funds a dedicated bioinformatics post embedded with the All Wales Medical Genomics Service (AWMGS) to evaluate the clinical utility of applying the latest genomic analysis software on patient data to improve the diagnostic rate.

For well over a decade, Wales Gene Park has collaborated with AWMGS to develop compute and data storage infrastructure to support genomic diagnostic services within the NHS. And now, working as partners within Genomics Partnership Wales, we continue to work together to further develop IT not only to support future healthcare, but also the medical research that will enhance that healthcare.

Through Wales Gene Park support, AWMGS now have their computer cluster (known as Wren) to handle the hundreds of clinical tests they perform each month. Alongside Wren we are in the process of building our Sêr Cymru IT that will serve as an equivalent resource to support cancer and rare genetic disease research. Together these two complementary IT resources are being brought together, to attract funding to create a combined resource that will allow research and commercial partners controlled access to genomic and other clinical data in a safe, secure environment.

Through specific use-cases, involving our Health and Care Research Wales partners (Wales Cancer Research Centre and Wales Cancer Bank), we are focusing in on specific data integration challenges that will help inform our combined data strategy. So for example, teaming up with Wales Cancer Bank we are working with AWMGS to sequence cancer tumour biopsy samples and link these data to histology images to create a combined data repository that will eventually support the development of new AI technology to better stratify cancer patients for more personalised treatments. Together with AWMGS, we are also working with Wales Cancer Research Centre partners to help develop an improved risk stratification model for identifying individuals at high-risk of colorectal cancer supporting the expansion of the existing CONSCOP2 trial to undertake mutational signature analyses to compare 'advanced polyps' to 'non-advanced polyps' to better identify those at a higher risk of colorectal cancer.

Case Study 2

Learning together. Training the next generation of Bioinformaticians.

A Bioinformatician uses technology, numerical and computer science skills to answer questions of biological significance. Clinical bioinformatics is a fast-growing crucial area of healthcare science.

There is a pressing need to ensure the education and training of the next generation of Bioinformaticians to meet the demands for skilled people to undertake these critical roles.

The demand for skilled individuals

As genomic technologies have advanced, the quality and amount of data that can be produced has increased. Being able to data manage, analyse and interpret the large genomic data sets with other sources of data is the role of a Bioinformatician. These roles are crucial to be able to understand and make the most of these data.

The role of a Bioinformatician is multidisciplinary, and requires developing skills in biology, maths/stats and computing, as well as professional values such as good communication, negotiation and leadership skills. This is a key and growing sector within health data science.

Developing future leaders

Providing a safe environment where students can try out bioinformatic techniques, make mistakes and learn from them together is offered at Cardiff University through the MSc Bioinformatics and MSc Bioinformatics and Genetic Epidemiology programmes. Our role is to inspire the next generation of Bioinformaticians, our future leaders.

A key highlight of these programmes is that learning outcomes are achieved through using real and current big data sets supplied by world leading experienced researchers and clinical academics. This offers opportunities to connect, learn and work with people, building on experience, and also developing a network of connections for the future.

“The support from the WGP has been particularly valuable for the MSc programmes we offer. Their commitment to student development is fantastic and is helping our students prepare for a successful future.”

Dr Marian Hamshere, Cardiff University.



Working together to deliver excellence

Wales Gene Park is one of our key health data science collaborators, offering data projects and supervisors to help mentor students as they learn.

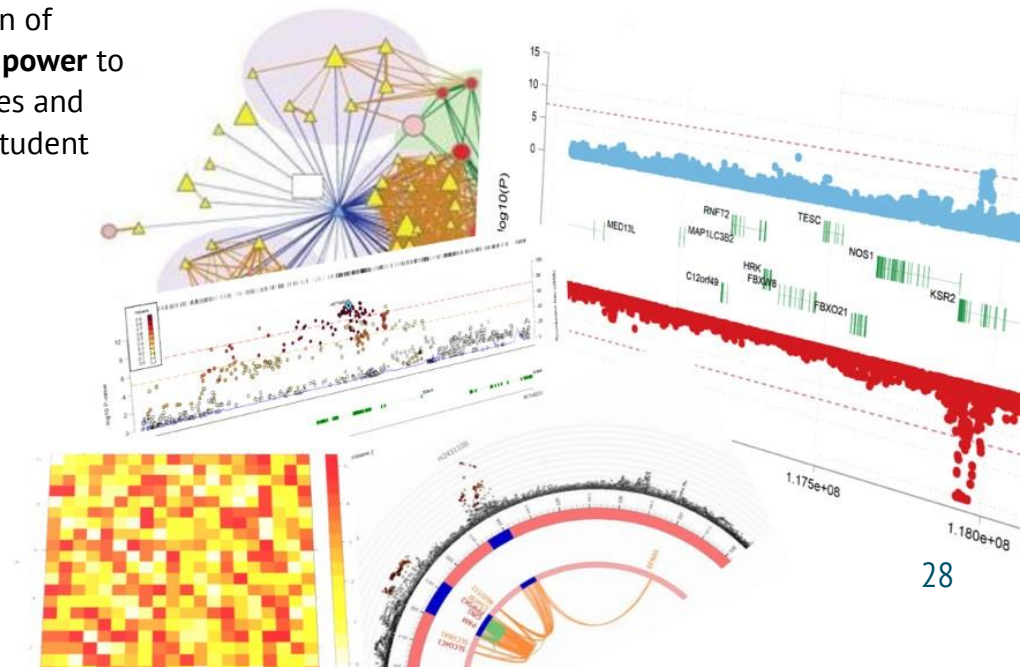
The real data sets are embedded in the taught and research elements of the programmes, offering publication opportunities to students from the start. Wales Gene Park provide **expertise and leadership** with the module lead for case studies, workshop delivery, and provision of **dedicated computing power** to enable minimal queues and shorter run times to student projects.

Wales Gene Park also contribute a transferable skills workshop providing support for job applications, as part of their commitment to help students develop professional skills useful outside of the programmes.

Student feedback demonstrates the value of this holistic approach to bioinformatics training, many highlighting the benefits of this engaging and inspiring format.

The success of the programme

100% of the 2018/19 cohort, and 75% of the 2019/20 cohort of students are working or furthering their studies in Bioinformatics. Two past students have gone on to work at Wales Gene Park.



Case Study 3

Working with commercial partners



The provision of flexible, bespoke Next Generation Sequencing (NGS) for research is one of WGP strengths. This agile way of working has supported the work of Prof Simon Reed (Cancer and Genetics, Cardiff University School of Medicine), contributing to the establishment of a biotech start-up company called Broken String Biosciences.

In simple terms, DNA damage is not good for the health of a cell, and hence the body in which the cell is found.

Cells have developed many complex mechanisms to deal with and fix DNA damage, processes known collectively as DNA repair. For over two decades, Prof Simon Reed's academic research has focused on understanding the functionality of DNA repair, and this has implications for many diseases, including cancer.

Prof. Reed has worked with WGP over the last few years, for support to utilise NGS technologies for his research.

By tweaking the standard NGS protocols, WGP were able to support Prof Reed and his research team to develop new protocols that used NGS technologies to map points of DNA damage within the genome.

The development of a quick, accurate, robust method to map breaks in the genome will directly enable safe development of new cell and gene therapies. Checking for and ruling out "off-target" damage following these cutting-edge therapeutic strategies will be essential to see them more widely used in the clinical setting.



<https://www.brokenstringbio.com>

Broken String Biosciences has received Illumina Accelerator funding and is operating at Illumina's European research HQ in Cambridge.

The company aims to advance therapeutic discovery and development by assessing the stability of the genome. The technology platform being produced combines novel sequencing techniques, bioinformatics analysis, and AI, to provide healthcare professionals with a data-driven approach for research developing the next generation of innovative medicines, including cell and gene therapies.

Broken String Biosciences aims to build a platform of genomics solutions for assessing genome stability (a measure of the amount of DNA damage that is occurring in a cell). The company are now building on their first technology known as INDUCE-seq.

Together, these novel DNA sequencing tools will be able to address an unmet need that exists within the life sciences, thereby removing barriers and providing methods that can assist with the development and regulation of the next generation of innovative medicines, including cell and gene therapies.



Prof Simon Reed, Cardiff University.
CSO Broken String Biosciences

Case Study 4

Genomic Data Integration

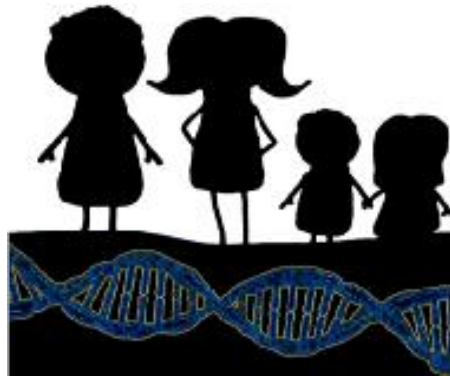
Linking epilepsy variants to routine data in the SAIL Databank: pathfinder study

The linking of Genomic Data with other datasets has the potential to provide a very valuable resource for researchers.

In the previous annual report, we summarised the 'Jedi' project – Genomic Data Integration (GeDI) into data safe havens. From that work, we published recommendations for a model to assess risk which has a flexible suite of controls to safeguard privacy and retain data utility for research. Since this project, we are working on a practical pathfinder study to link routine data held in the SAIL Databank with genomic data for an epilepsy study.

The aim of the pathfinder project.

This project is looking to link genetic data (gene-variant datasets) from next-generation sequencing to health records within the SAIL databank. It is a pathfinder study because the focus of the work is to explore appropriate methods and linkage processes. The project will develop guidelines and protocols for others wishing to integrate genetic data with routine data. As a small study (N=111), it is unlikely to produce properly meaningful results when assessing the potential association between genetic variants and clinical outcomes, but this proof of concept will provide the foundations for others to follow.



Progress of the pathfinder project

The genetic datasets were in the form known as Variant Call Format (VCF) files. These were derived from samples donated by Welsh patients to the Swansea Neurology Biobank. The DNA was sequenced as part of the Epi25 collaboration (<http://epi-25.org/epi25-cohorts>) and all patients included in the study consented for their genetic data to be uploaded to SAIL. The need for consent to linkage is important to note since the data was collected for research.

Following annotation of the VCFs, we linked the resulting data with GP and hospital records in SAIL. We used the linked data to examine the association between genetic variants and epilepsy outcomes. Via this linked data we were able to explore questions such as the genetic burden and epilepsy severity as measured by unscheduled hospital admissions and by the use of anti-seizure medications. We will be reporting on the data preparation, linkage methods and example questions in a paper submitted for peer-review publication.

Next steps

The experience and learning from this work together with that of the Jedi study will be used to develop rules of engagement for integrating genetic data with routine data in SAIL, with relevance for trusted research environments more widely.



Beata Fonferko-Shadrach

Data Science Researcher, Swansea University

**For more information contact the project lead Professor Kerina Jones
Project Lead, Swansea University
(k.h.jones@swansea.ac.uk).**

Case Study 5

Genomics Cafés

Why a Genomics Café?

In July 2017, Welsh Government published its *Genomics for Precision Medicine strategy* which identified key actions to ensure people in Wales benefit from the advances in genomic medicine. As a result, Genomics Partnership Wales (GPW) was set up to deliver this vision.



To help shape progress in this area, it is key that patients and the public are involved in these advances. As part of GPW's programme of engagement, Wales Gene Park has established a new Public Genomics Café initiative, aimed at engaging with the public and those affected by rare and genetic conditions about genomics, in an informal environment.



Café Culture

Since their inception in summer 2019 the cafés have been held in-person at public-friendly, accessible venues, such as coffee shops, throughout Wales. The cafés are free, open to all, and aim to be a friendly and engaging environment through which information about genomics, and its relevance to health and medicine, can be shared and discussed.

Café Menu

Events comprise short, relaxed talks from guest speakers on relevant topics, who include:

- Researchers, who can engage with attendees about their research, signpost to participation or involvement opportunities and seek feedback on their work.
- Health professionals, such as Genetic Counsellors or doctors from the All-Wales Medical Genomics Service.
- Those affected by rare or genetic conditions, who share their personal experiences.

Congratulations on a brilliant café this morning. Fascinating and hugely instructive. I only wish that all three presentations and the associated discussions could have been seen by everyone. Can't wait for your next session!

Café attendee

Café Connections

Based on informal discussions with attendees, networking has led to useful conversations and interactions - for example, those affected by rare conditions being signposted to information or services through speaking with health professionals; researchers receiving feedback on their work; members of the rare disease community making new connections. Relaxed Q & A sessions have also been popular.

Meeting others face-to-face in a relaxed environment has been so beneficial. Despite having different rare conditions, so many common concerns and issues. Great to have the opportunity to hear and share experiences and the good practices that can make life easier and more manageable.

Rare Disease Parent Carer & Advocate

Virtual Cafés

Since May 2020 events have been virtual with themed cafés, online engagement and e-networking (as and when requested by participants). Translation to a virtual platform has been successful, with a positive effect on attendee numbers and broader audiences from across Wales and beyond. Six virtual Genomics Cafés have been held - reaching over 420 people.



Café take-aways

Very positive feedback has been received on the cafés to date with attendees reporting that they are enjoying the format and content. Participants also get in touch to request follow-up information and suggest topics.

I have been to several of the cafés to discuss my research. Members have given really useful feedback on my work, as well as offered to participate in my study. I have had the opportunity to meet people I otherwise would not have, learned a lot about other work being done in the community and also been able to gain better insight about some of the challenges facing people living with a rare condition. I plan to keep attending for as long as they're being held!"

Genomics Café Speaker

Café Highlights

- Genomics Cafés were highlighted as a case study in the UK Government's Genome UK - The future of healthcare policy paper, which sets out the vision to extend the UK's leadership in genomic healthcare and research.
- The launch in October 2020 of a new Young People's Genomics Café aimed at the 16 to 25 age group. Two events have been held to date, attracting over 225 attendees.
- Opportunities for café participants affected by rare or genetic conditions to share their experiences during educational sessions for healthcare students.
- Co-organisation of a specialist Genomics Café focusing on haematology (taking place in June 2021) with the patient organisation *Friends of Cymru Sickle Cell & Thalassaemia* and specialist health professionals.

Plans for future Genomics Cafés include:

- Guest slots from other Welsh Government work programmes such as Healthcare Sciences and Advanced Therapies Wales.
- Involving stakeholders including patients and the public, to co-organise and co-host future cafés.
- Exploring a blended approach of online and face-to-face events, with the aim of making them accessible to a broad audience.
- Scope for future cafés to include activities such as focus groups, consultation responses and targeted discussions around topics relevant to the Genomics Partnership Wales programme.

If you are interested in finding out more about the Genomics Cafés, please get in touch:

walesgenepark@cardiff.ac.uk

Looking Forward

Supporting genomic research in Wales

Wales Gene Park is an integral partner within Genomics Partnership Wales and we recognize collaboration is key to support genomic research in Wales: Collaboration with the NHS, with industry, and with other research organisations funded by Health and Care Research Wales and beyond.

Collaboration is essential because the resources needed to enable and support genomic research are expensive and require continual updating, and because the key data sets that we need to link together to support medical research are curated by numerous data controllers across academia and within the NHS.

This is why Wales Gene Park is working ever more closely with our Health and Care Research Wales funded partners, such as Wales Cancer Bank and Wales Cancer Research Centre, and Genomics Partnership Wales partners, to focus in on the key enablers for translating medical big data into patient benefits.

There are several key enablers we need to be focusing on that Wales Gene Park can play a role in supporting.

Big data: If we are to realise the benefits of Artificial Intelligence in Healthcare, we increasingly need to collect data at scale. Through our work as part of Genomics Partnership Wales, we are continuing to build the infrastructure needed to generate genomic data at scale and the IT wherewithal needed to store and analyse that data.

Data linkage: To be truly useful for research and healthcare, genomic information needs to be linked to other data types. Through our close collaboration with the SAIL Databank we continue to explore ways of safely linking genomic data to other anonymised epidemiological information for population research benefit. And now, as we with our partners increasingly work with the NHS Wales National Data Resource, we are expanding opportunities for data linkage.

Data access: Wales Gene Park has long recognised the importance of providing researchers with safe, secure access to genomic data in a way that enables research but does not compromise the confidentiality of research participants and patients. Working with the SAIL Databank provides us with the opportunity to inform a coordinated data access strategy across our partnerships, drawing on existing expertise within other Health and Care Research Wales organisations to inform our collective data strategy.

Data sharing: We need common, shared standards among partners so that when we do share data we do so in a way that is consistent and allows us to build a coherent federated data infrastructure to benefit Wales. Wales Gene Park is working with Wales Cancer Research Centre and others to ensure we adopt the right standards such as those genomic standards promoted by the Global Alliance for Genomics and Health (GA4GH).

IT: Through initiatives such as the Sêr Cymru IT project, the AWMGS Wren computer cluster, and the work of Genomics Partnership Wales, Wales Gene Park is helping to drive the development of the right IT infrastructures needed to support genomic data generation and analysis in Wales and to do so in a way that is sustainable to support research well into the future.

Expertise: Wales Gene Park has always been about attracting and retaining the right skill sets needed to support genomic medicine in Wales. We continue to provide specialist laboratory and bioinformatics staff to support genomics research and we are working with other Health and Care Research Wales partners to build on this nucleus of expertise. Through our expertise and leadership, we are actively supporting Wales Cancer Research Centre build their data scientist workforce, and we continue to fund staff embedded within the AWMGS and the SAIL databank.

www.walesgenepark.cardiff.ac.uk

Acknowledgements

We've heard it said, on more than one occasion, that Wales Gene Park achieves a lot for a small team, and this is in part due to the support received from many senior academics, Cardiff University colleagues with NGS expertise, the university management and NGS staff who continue to support Wales Gene Park. We thank them all for their invaluable help. We further thank all members of the strategic advisory group and our patient representative for their continued and excellent support and advice.

Conclusion

In a year like no other, the application of genomic technologies has shone through. The work of Genomics Partnership Wales colleagues, especially that of Public Health Wales Pathogens Genomics Unit (PenGu) has epitomised the versatility and value of genome sequencing for public health. Similarly, the application of whole genome sequencing analysis of seriously ill children and their parents by the All Wales Medical Genomics Unit has demonstrated the power of these techniques to deliver faster diagnosis for some children, more timely treatments and better outcomes. The outputs of the momentum spoken of in previous year's reports are being seen in abundance.

As an infrastructure support group, Wales Gene Park comprises a team of genomic technologists, bioinformaticians and education and engagement practitioners.

The Education and Engagement team have again delivered a large programme of events for a range of audiences, drawing on expertise in the Welsh Higher Education Institutions and NHS. This feat was achieved under extraordinary circumstances, with all in-person events being cancelled and replaced by virtual meetings. We very much hope that we will be able to return to operate with a hybrid approach going forward, keeping some of the advantages of virtual work, but incorporating in person events soon. Through our endeavours we work to ensure the Welsh Public are informed and can make the most of the advances of genomic technologies in health care.

The laboratory, bioinformatics and data analysis activities within Wales Gene Park have continued to provide support for a range of research in Wales. Past endeavours have been incorporated into 36 publications within the reporting period, advancing scientific knowledge in a range of scientific fields.

Investment in dedicated IT infrastructure for genomic research will go some way to ensuring the provision of high-calibre research using genomic data. However, as the rate of data generation increases, and the ability to link genomic data with other data sets comes to fruition, the continued investment in IT infrastructure will need to continue at pace. Collaborations will be key to see this materialise.

Glossary

1-9

3G Third Generation

A

AI Artificial Intelligence

ARCCA Advanced Research Computing at Cardiff

AWMGS All Wales Medical Genomics Services

C

CONSCOP2 A randomised controlled trial of contrast enhanced colonoscopy in the reduction of right sided bowel cancer

COVID-19 Coronavirus disease

CPD Continuing Professional Development

CRISPR Clustered Regularly Interspersed Short Palindromic Repeats

D

DHSC Department of Health and Social Care

DNA Deoxyribonucleic acid

E

ES Embryonic Stem

G

GA4GH Global Alliance for Genomics and Health

GB Great Britain

GeDI Genomic Data Integration

GPW Genomics Partnership Wales

I

iPSCs induced Pluripotent Stem Cells

M

MDT Multidisciplinary Team

MS Members of the Senedd

N

NGS	Next Generation Sequencing
NHS	National Health Service
NIHR	National Institute for Health Research
NSHS	National School of Healthcare Science

P

PenGU	Public Health Wales Pathogens Genomics Unit
PPI	Patient and Public Involvement

Q

QC	Quality Control
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R

RDIG	Rare Disease Implementation Group
RNA	Ribonucleic acid

S

SAG	Strategic Advisory Group
SAIL	Secure Anonymised Information Linkage
SCW	Super Computing Wales
SWAN	Syndromes Without a Name

T

TB	Terabytes
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U

UK	United Kingdom
UKCRN	UK Clinical Research Network
USA	United States of America

V

VCF	Variant Call Format
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W

WCB	Wales Cancer Bank
WCRC	Wales Cancer Research Centre
WHSSC	Welsh Health Specialised Services Committee
WGP	Wales Gene Park

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