ANNUAL REPORT
2021-2022
WALES GENE PARK

2021 - 2022
Annual report
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“Harnessing Genetics and Genomics to advance Research, Healthcare, Education and Innovation”

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Welcome

Annual Report 2022

Welcome to the Wales Gene Park annual report which summarises Wales Gene Park activities during the period 1st April 2021-2022.

The past two years have been difficult for everyone. The Coronavirus pandemic presented numerous challenges and made us all re-assess how we live and work. New ways of working were adopted, and some have been so successful that they will be maintained for the future. During the COVID-19 era Wales Gene Park has continued to operate and achieve its objectives.

This annual report captures the value and impact of the broad portfolio of work undertaken by the Wales Gene Park. The common purpose of these activities is to ensure that the full benefits of genomics are realised in Wales. To achieve this, Wales Gene Park is always looking at new collaborations and ways of delivering its objectives successfully. Wales Gene Park is an integral part of Genomics Partnership Wales and works with the other partners to achieve the best possible outcome for the people of Wales.

Genomics is a rapidly developing field and Wales Gene Park is looking forward to the opportunities and challenges ahead and celebrating its 20th Anniversary next year.

Angela Burgess
Education & Engagement Lead/Co-Director.
Foreword

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

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

Genomic science is an essential cornerstone of biomedical research and has a growing impact on healthcare. The work of the Wales Gene Park ranges from technical developments in the laboratory to supporting the development of public policy by Welsh Government, but the common purpose of these diverse activities is to ensure that the full benefits of genomics are realised in Wales.

This year the Wales Gene Park has generated sequencing data for 55 genomic projects, provided bioinformatics support for 67 projects and supported external research grants bringing in over £4.3M to Wales. Wales Gene Park has provided support to researchers working on cancer, immunology, rare disease, and others.

We involve patients and the public in the planning and execution of research and in service and policy development through an extensive programme of engagement. We promote understanding of the opportunities and challenges raised by genomics among health professionals, schools and colleges and the public through a wide portfolio of education activities. We are receptive to ideas, questions, and comments. To get in touch or to find out more about us please do visit our website at www.walesgenepark.cardiff.ac.uk.
A word from our Patient & Public Representatives

"It’s fantastic to see the breadth and depth of work that Wales Gene Park carries out and great to have the chance to be involved in it. It is really reassuring and refreshing that Wales Gene Park puts the public and patients at the heart of everything it does. The 2209 members of the public/patients, who attended the 49 events during the last year, show that there is considerable interest in and a good appetite for the work carried out by Wales Gene Park. I’m really looking forward to continued involvement in the future”.

Mr Nathan Davies, Patient Representative

It is amazing to see the huge amount of work that the Wales Gene Park undertakes, and I am pleased to be part of it. You could say that it is “Patient infused” in all of its projects.

Mr Alan Thomas, Patient Representative
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 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.

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

https://www.walesgenepark.cardiff.ac.uk/our-teams/
Who’s Who and Key Partners 2021-2022

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  
• Cwm Taf Morgannwg University Health Board  
• Swansea Bay University HealthBoard  
• St Marks, London,  
• Partners within a musketeers’ memorandum study

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, 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 Interaction Lead

Patient Representative
• Mr Alan Thomas  
• Mr Nathan Davies

Strategic Advisory Group
• Chair: Professor Colin Dayan, School of Medicine, Cardiff University  
• Mrs Emma Hughes, 3rd Sector Representative and WGP PPI Lead  
• Dr Rob Orford, CSO Health, Welsh Government  
• Dr Dee Riple, 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  
• Dr Clive Morgan, Cardiff and Vale University Health Board  
• Sian Morgan, Head of Laboratory at the NHS All Wales Medical Genomics Service Laboratory  
• Michaela John, Genomics Partnership Wales.  
• Dr Mark Bale, Head of Science Partnerships, Genomics England  
• Dr Francis Sansbury, Health and Care Research Wales Speciality Lead for Genomics and Rare Diseases
Wales Gene Park is funded by Welsh Government through Health and Care Research Wales to enable genomic research in Wales. During the 2021-2022 reporting period we supported research via two work packages.

Work Package 1
Genomics 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.
Development of IT infrastructure that enables research using genomic data.

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

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 enabled sequencing access for 55 projects, 55% 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, Sepsis, neurodegeneration and Alzheimer’s projects.

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, most recently through the recently purchased Sêr Cymru IT infrastructure and we are now leading a consortium of Health and Care Research Wales -funded organisations and other partners to improve Trusted Research Environment capability in Wales for cancer and rare genetic. Our focus is on unlocking the potential of healthcare genomics data for translational research in a way that is safe, secure, and trustworthy.
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.

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 25 new patient consents and banking 39 samples from patients with cancer or rare diseases, we have supported four laboratory-based research projects that aim to improve future NHS diagnostics.

In addition to this activity, Wales Gene Park worked to lay the foundations needed for several clinical trial research studies. The details of this activity are highlighted in case study 2.

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. Using human induced pluripotent stem cells (iPSCs), CRISPR (Clustered Regularly Interspersed Short Palindromic Repeats) genome editing, cell differentiation and organoid technologies, we have supported 12 research groups on 15 projects. Projects have included the generation of novel human cell models to study genetic risk factors that influence neurodegeneration and neuroinflammation in Alzheimer’s disease, Huntington’s disease, Glioma, neuropathy of the eye and sensory neuron function in osteoarthritis. This work has equipped the research groups with new tools that can be used to take their research forward.

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 86 events organised by Wales Gene Park, we have reached:
- 1113 Health Professionals: 25 events
- 655 Schools/Colleges students/teachers: 12 events
- 2209 Public/Patients: 49 events

Case study 4 shines the spotlight on the Genomics Showcase event, that the Wales Gene Park Education and Engagement team helped deliver. This event has received feedback that has been overwhelmingly positive, with calls for future events to maintain elements of the virtual format.

Enabling patient involvement in research
Wales Gene Park facilitated and advertised opportunities for patient and public involvement in research throughout 2021/2022. Opportunities were circulated to our rare disease patient network, as well as the Genomics Partnership Wales Patient and Public Sounding Board. Attendees at events such as the Genomics Cafes were also made aware of opportunities for involvement. In total, 31 involvement opportunities were advertised with 263 individuals taking them up during the reporting period. One example is the ‘Unique You Project’, which involved public workshops with members of the cancer community in Wales to discuss and shape the development of tools to explain topics relating to cancer, genomics, and personalised medicine to the public and patients.
Core Metrics
Reporting period: 2021/2022

Health and Care Research Wales infrastructure award to the group

- Direct funding awarded: £734K
- Jobs created through direct funding

Grants won during reporting period

<table>
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<th>Grants won</th>
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<th>Group collaborating</th>
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</tr>
<tr>
<td>Additional jobs created for group</td>
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</tr>
</tbody>
</table>

- Number of publications: 32
- Number of public engagement events: 80
- Number of public involvement opportunities: 31
Supporting research across Wales and beyond
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 SAIL databank (Swansea)
- Contribution to healthcare professional education
- Derivation and Growing human iPS 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

Purpose and Progress

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

- A Next Generation Sequencing (NGS) laboratory (55 projects supported in the last year)
- Bioinformatics provision (67 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

More details about the work undertaken to enable clinical research trials can be found in case study 2.

Genomic data integration into the SAIL databank

The Wales Gene Park award supports a pathfinder project that is looking to develop the methods needed to integrate genomic data into the SAIL databank. This project is underpinned by data derived from samples donated by Welsh patients to the Swansea Neurology Biobank, which were sequenced as part of the Epi25 collaboration (http://epi-25.org/epi25-cohorts). The variation within the genetic data is stored in a file type known as Variant Call Format (VCF) files. Information from epilepsy clinic letters has been successfully extracted using our natural language processing application (ExECT). This information has been linked to the genetic data and health records within the SAIL databank.

Several methodological challenges have arisen from working with data in different formats. This novel work has uncovered and resolved many of these issues, and allowed an investigation to be undertaken to look for associations between the genetic variation and the ability to control epilepsy symptoms. Epilepsy control was measured by the surrogate markers of anti-seizure medication polytherapy, unscheduled hospital admissions and by the seizure frequency information extracted with ExECT. This work has been accepted for presentation at the International League Against Epilepsy (ILAE) 14th European Epilepsy Congress, July 2022. The description and solutions to these methodological challenges, lays the foundations for others working with data in differing formats to link genomic data with structured data within the SAIL databank.
Enhancing understanding of genetics and genomics throughout Wales
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:

Community Group talks

Thirty DNA and genetics/genomics-related talks were delivered to community-based organisations throughout Wales.

Genetics & Genomics for the 3rd Generation (3G) public conferences

The 6th Annual 3G conference took place virtually. It is specifically aimed at the over 50 age group, but open to any members of the public with an interest in DNA, genetics, genomics, and associated topics. Over 100 attendees joined via Zoom to enjoy talks ranging from ‘How your genes can affect the medicines you take’ to ‘Nutrigenomics’.

Public Lectures

Wales Gene Park’s public lectures are aimed at engaging with a lay audience about genetics and genomics-related subjects. One of this year’s lectures, held as part of Cardiff University School of Medicine’s Science in Health Public Lecture Series, has been viewed again over 9000 times.

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

Genomics Cafes

Public Genomics Cafés, organised as part of the Genomics Partnership Wales engagement programme that Wales Gene Park oversees, include short talks about genomics relating to health and medicine.

Since April 2021, seven public cafes have taken place attended by more than 420 people. One of these was a haematology themed café co-produced with the patient organisation Friends of Cymru Sickle Cell & Thalassaemia. Three Young People’s Genomics Cafés, aimed at the 16 to 25 age group, were attended by over 100 people.
Schools and Colleges

Activities for Schools and Colleges continued to be affected severely by the COVID-19 pandemic. Despite this, WGP delivered a programme of engagement including Teachers’ Genetics Network newsletters, a range of virtual talks, and online content for several events and Science Festivals.

School & College Genetics and Genomics Roadshow

Now in its 12th year, in 2021/22 the Genetics and Genomics Roadshow went virtual for the first time. The roadshow offers free talks to year 12/13 students, with experts speaking on DNA, genetics & genomics-related topics and discussing careers. Despite the significant COVID-19 disruption, a series of events was delivered throughout Wales attended by over 400 year 12/13 students.

Tremolo – a podcast drama about early-onset familial Alzheimer’s disease for post-16 students and the wider public

Tremolo is an exciting new project with Illumine Theatre and Theatre Genedlaethol Cymru. Aimed primarily at audiences aged 16+, Tremolo – from playwright Lisa Parry – is a thought-provoking, bilingual podcast drama exploring the impact of a diagnosis of early-onset familial Alzheimer’s disease (eFAD) and some of the issues around genetic testing. It is accompanied by a bilingual educational pack for teachers, aimed at year 12/13 students. The project received funding from The Genetics Society, the Adelphi Genetics Forum and Theatr Genedalethol and is available to listen and download via platforms including Spotify, Apple Podcasts and AM. Further details about Tremolo can be found in the accompanying Case Study 5.

Health Professionals and Researchers

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

A Machine Learning Applications in Genomics Webinar held in partnership with SuperComputing Wales; the COVID-19 Genomics UK Consortium Welsh Hub: Pathogen Genomics Unit Celebration of Endeavour and Achievement, held to mark the end of routine SARS-COV-2 sequencing and celebrate the achievement; the annual SCE Clinical Genetics Revision Course, hosted by the AWMSG, held for doctors training and specialising in this area of medicine.

Education sessions

A series of Living with Genetic Conditions sessions – focusing on the patient experience - were delivered to health professional students at higher education institutes throughout Wales. Educational sessions were also delivered to post-graduate students and researchers.

WGP also continued to engage health professionals and researchers with an interest in genetics and genomics through its new Professionals’ Genetic Network and associated newsletters.

“Really enjoyed this session! The ethical scenarios really made me consider different perspectives of both arguments especially with the incidental findings.”

“The students had their eyes opened to what an A level Biology course could lead on to. The talks were very interesting, and students were positive about the session.”
**Genomics Partnership Wales Genomics Roadshow talks**

Sessions aimed at engaging with health professionals about advances in genomic medicine in Wales, including Welsh Government’s *Genomics for Precision Medicine* strategy, were delivered throughout Welsh Health Boards.

**Supporting Research Engagement**

WGP’s Education and Engagement team is partner in the Unique You - How your genome defines your healthcare project being led by Cardiff University researchers. The project aims to work with public partners to co-design engaging resources to inform and raise awareness of genomics and precision medicine. WGP is supporting the project in several ways, including using its networks to connect researchers with the public, supporting engagement workshops, and helping to trial new resources at public events. This project provides a good example of how WGP can facilitate and support researchers in undertaking engagement.

**Patients & Families**

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

**Annual Rare Disease Patient Network meeting**

A key priority for this year’s annual meeting was involving the rare disease community in identifying key priorities to include in a draft Action Plan (overseen by the Rare Disease Implementation Group, RDIG) to implement the UK Rare Diseases Framework (2021) in Wales. In 2021, two consultation workshops (one involving network members at the annual meeting and the other, the Genomics Partnership Wales Patient and Public Sounding Board) were delivered by Genetic Alliance UK and Dr Graham Shortland (Chair, RDIG), to better understand key priority areas. The workshops engaged with over 50 people affected by rare genetic and undiagnosed conditions across Wales and the outcome was a series of recommendations in relation to the four priority areas of the UK Rare Diseases Framework. The underpinning themes of the Framework were incorporated across each priority area of the Welsh Action Plan, and a report was presented to the group responsible for producing the Welsh Plan.

**Rare Disease Nurse Network collaboration**

Working in collaboration with Rare Disease Nurse Network, Genetic Alliance UK and the Chair of RDIG, WGP worked to involve patients and carers in the development of a survey exploring nurses’ experiences of care coordination in relation to rare conditions in Wales. Survey responses supported the development of a Syndromes Without a Name (SWAN) clinic in Wales.

**Annual Rare Disease Day Reception 2022 & launch of ‘Good Diagnosis’ report**

The theme of this year’s annual Rare Disease Day parliamentary reception – which aims to improve stakeholder awareness through patient experiences - was a ‘Good Diagnosis’. In December 2021, Genetic Alliance UK set out to better understand people’s experience of diagnosis and to identify what matters most to people with a rare condition when they are on their diagnosis journey.

The findings were launched at the Rare Disease Day reception and final report can be found here: [https://geneticalliance.org.uk/wp-content/uploads/2022/02/Rare-Disease-UK-Good-Diagnosis-Report-2022-Final.pdf](https://geneticalliance.org.uk/wp-content/uploads/2022/02/Rare-Disease-UK-Good-Diagnosis-Report-2022-Final.pdf)
Genomics Partnership Wales Patient & Public Sounding Board Consultations
Wales Gene Park worked with Genomics Partnership Wales through its PPI Sounding Board to provide input and feedback on a range of genomics-related topics including targeting health professionals and canvassing public understanding around genomics. WGP also supported 4 further consultation events throughout this reporting period, to ensure the patient voice was key to the implementation of the Genomics for Precision Medicine Strategy in Wales.

Cross Party Group Meetings
At the start of the Senedd's sixth term, Genetic Alliance UK held a parliamentary briefing to support understanding of rare, genetic, and undiagnosed conditions amongst Senedd Members. The Cross-Party Group has now been re-established under new leadership and with cross party support and the group has been supporting the development of a Welsh Action Plan by highlighting key areas of focus.

Involvement in Research
Wales Gene Park facilitated and advertised opportunities for patient and public involvement in research and other activities throughout 2021/2022. Opportunities were circulated to WGP’s Rare Disease patient and public networks as well as the Patient and Public Sounding Board and attendees at events such as Genomics Cafes. In total, 31 involvement opportunities were advertised with 263 individuals taking them up during the reporting period.

Facilitated by WGP, one of its Rare Disease Patient Network members worked with the Health and Care Research Wales Communications team, to produce a news item for Rare Disease Day 2022 to raise awareness of rare conditions and highlight the importance of research in this area:


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:

- Building on the investment of the Sêr Cymru funded IT infrastructure, Wales Gene Park leads a consortium of Health and Care Research Wales-funded organisations and other partners, to develop an integrated digital strategy for Wales that most recently has included funding secured to develop improved Trusted Research Environment services in Wales. Further details of this project are outlined in Case Study 1.

- Impact from research can only be realised when the foundations needed to enable research to happen, are themselves in place. The Wales Gene Park bioinformatics team have continued to work with colleagues to establish the foundations needed for several clinical trial research projects. Case Study 2 outlines the progress made in this area during the reporting period.

- Case Study 3 highlights the work of the Wales Gene Park Research Co-ordinator, whose work supports several research studies focused on rare genetic diseases. The most recent findings from one of these studies, that facilitated a new diagnosis to be made for patients in Wales, is described in detail.

- The Genomics Showcase was held as a virtual event in May 2021, with over 1200 people registering. The content was available for a further 30 days after the live event. The highlights and overview of the Showcase are provided in Case Study 4.

- Case Study 5 outlines a very creative approach of communicating with the public about genetics, genomics and scientific topics. The podcast drama Tremolo explores the impacts of a diagnosis of early-onset Familial Alzheimer’s Disease (eFAD) and some of the socio-ethical issues around genetic testing.
Case Study 1
Developing Wales’s Digital Infrastructure for Genomic Research

Working with Health and Care Research Wales partners and the Experimental Cancer Medicine Centre (ECMC) in Cardiff, Wales Gene Park has secured industry funding for a 2-year project to develop a Trusted Research Environment (TRE) capacity for cancer & rare genetic disease research in Wales.

The project will be delivered in close partnership with the SAIL Databank and the National Data Resource programme (NDR) and informed by the Wales Cancer Research Strategy, (CReSt).

This case study describes Wales Gene Park’s success in leading academia, healthcare, and industry to help develop Wales’ digital infrastructure for genomics research. Working with key strategic partners that include other Health and Care Research Wales funded organisations, and the AWMGS, Wales Gene Park has over the last year established the Sêr Cymru IT infrastructure and through this work has now secured £560K funding from a commercial partner to expand on this work to evolve Wales Trusted Research Environment services through the Wales Cancer TRE Project.

Laying digital foundations with the Sêr Cymru IT

The Sêr Cymru IT is high-performance computing and digital storage designed to support cancer research. Built with an £277K investment from the Sêr Cymru II Infrastructure Fund (WEFO ERDF Programme 80762), the Sêr Cymru IT now provides dedicated, resilient, and above all secure computing and data storage to Health and Care Research Wales partners and key Cardiff University cancer groups. Sêr Cymru IT is now run by Wales Gene Park on behalf of those partners to support existing research and pilot new ideas. The Sêr Cymru IT is a steppingstone in the evolution of Wales’ digital solution for genomics research and will form part of the digital infrastructure that will support Genomics Partnership Wales at the new Cardiff Edge development at Coryton.

Over the last reporting year, Wales Gene Park have worked with partners to use the Sêr Cymru IT to develop pilot projects (for example, Wales Cancer Bank’s collaboration with Panakeia) and develop further projects (for example, supporting the recent successful bid to establish an EPSRC Interdisciplinary Doctoral Training Hub for precision oncology, aligning with our genomics training ambitions).
Building on those foundations with the Wales Cancer TRE Project

Sêr Cymru IT has now led to work with partners and especially the Wales Cancer Research Centre that has secured industrial funding to develop the Wales Cancer TRE Project, a two-year piece of work designed to inform the development of Trusted Research Environment services for cancer and rare genetic disease in Wales.

Trusted Research Environments, or TREs, are increasingly used by academia, healthcare, and industry as a way of providing controlled researcher access to healthcare and other sensitive data. TREs provide safe havens in which researchers, appropriately vetted and trained, are granted access to data under a strict governance regime that controls what can be done with the data and what data can leave. A well-designed TRE both enables research whilst protecting the interests of the patients and public who have consented their data to be used in research.

The SAIL (Secure Anonymised Information Linkage) Databank is Wales’ databank of anonymised data about the population of Wales. As such, it is a leading TRE in the UK and through Health Data Research UK (HDR-UK) it has informed and guided the development of TREs that is now informing long-term UK Governments digital strategy as exemplified by the recent Goldacre Review: “Better, broader, safer: using health data for research and analysis”, independent report led by Professor Ben Goldacre.

The Wales Cancer TRE Project has been established to work with SAIL to enhance Wales’ TRE services for cancer research to unlock the research and development potential of Wales’ rich cancer and rare genetic disease data resources and do this in a way that is safe, secure, and trustworthy.

Working with the SAIL Databank, and informed by Wales research strategy for cancer, the CReSt strategy, the Wales Cancer TRE Project will align the shared digital ambitions of multiple Health and Care Research Wales-funded bodies into a single unified plan around shared digital infrastructure that complements and informs the Genomics Partnership Wales digital strategy.

The Wales Cancer TRE Project is led by research need

The Wales Cancer TRE Project is already up and running and partnering with multiple cancer research programmes to develop practical ways of improving secure, safe access to health and care data for cancer translational research with a particular focus on improving access and use of genomic data for precision medicine benefit.

Through these programmes of research, that include the development of early cancer detection and diagnosis technologies, the implementation of ground-breaking spatial genomics, and decision support system technology to support cancer treatment, the Wales Cancer TRE Project is identifying the cancer TRE services required in Wales. This will help us to unlock the potential of Wales’ rich data resources and provide the resources to work with SAIL as well as the National Data Resource, NDR, to enable those services.

Over the coming year we expect to see progress in the following areas:

- How to better leverage genomics to support early colorectal cancer detection.
- How to exploit spatial genomics to support translational prostate research.
- How to work with the Wales Cancer Bank to align genomic with image data to aid lung and breast cancer diagnosis.
- How to work with the Cardiff Experimental Cancer Medicine Centre (ECMC) to use genomics to improve patient recruitment to clinical trials.
- How to work with the Marie Curie Research Centre to develop their SMART-MDT strategy of an AI-driven decision support system to improve treatment decisions through cancer multi-disciplinary team meetings.
Case Study 2
Supporting Clinical Trial Research

Identifying cancer patients for a clinical trial
A cancer patient in Wales, who had not reacted well to the standard cancer therapy, was facing a situation of having very limited treatment options available to them. They had received a genomic test as part of their routine NHS management plan. The WGP bioinformatician working with the clinical genomics diagnostic lab in Wales (All Wales Medical Genomics Service, AWMGS) and the NHS clinical oncology team, analysed stored genomic data to identify patients potentially eligible to participate in a clinical trial for a new targeted cancer therapy. The new drug being tested in the clinical trial specifically targets a rare genetic variant present in less than 5% of all colorectal cancers. The analysis of the patient’s cancer genomic data provided a new avenue of treatment. Just before Christmas 2021, they were able to be recruited to the clinical trial. Follow up tests showed that the cancer had reduced in size in response to this new trial treatment. This is an excellent example of personalised treatment and highlights the value of genomics in improving care for cancer patients.
Using NHS data to advance research into Acute Myeloid Leukaemia (AML)

Another example of Wales Gene Park helping clinical trials researchers to utilise routine NHS data has come from working with the AML trials team. In this project, the AML trials team were looking to access an NHS held dataset for research consented trials patients. The AML trials team wanted to use this data to compare and confirm findings from an ongoing research project using a larger cohort of patients. This work is ongoing. While the request to access the data may appear straightforward, the data security and governance requirements for working with NHS data are not straightforward. They are purposefully, and understandably constrained. The WGP research bioinformatician embedded within the NHS AWMGS team, worked to facilitate the exchange of key data in both directions, between the NHS and the AML trials team, whilst negotiating the complex requirements for data security and governance. This project example highlights the need for an individual with the appropriate expertise, competencies, and workload capacity to be embedded in the NHS setting, to allow researchers to capitalise on clinical data for research.

Identifying Nuclear Export Signals for an international AML trail

Using his expertise and the WGP servers and dedicated computer, Dr Peter Giles, Wales Gene Park bioinformaticians has managed to recreate a lost online analysis portal, critical for the ongoing analysis within the international NCRI Acute Myeloid Leukaemia clinical trial. The trial utilised a tool from the Technical University of Denmark to assess nuclear export signals observed in novel mutations in the NPM1 gene which is an important molecular marker for Measurable Residual Disease. Unfortunately, this tool was discontinued by the authors and host institution leaving a key gap in the trials process. Following a review of options, Dr Giles worked with trial leads to obtain the code for the lost software tool and worked to get it running on more modern compute platforms. Dr Giles also creating a web-interface, allowing non-experts a route to use the command line tool. This work has plugged a gap that the AML trials team were facing, and in doing so allows the team to continue to provide this analysis for the ongoing clinical trial. [Image of the tool outputs is shown]
Case Study 3

Enabling Diagnosis through Research

The Wales Gene Park research co-ordinator plays a critical role supporting numerous rare disease research projects that have a genetic and genomic focus. These studies are registered as part of the Health and Care Research Wales Portfolio. Here we highlight the recent findings from one of these studies, that has enabled patients to receive a genetic diagnosis for their rare disease.

Project background

The Research study entitled “Genetic mechanisms in polyposis of the bowel” aims to identify the genetic changes that give rise to polyps (small growths) in the intestine.

Several rare disorders such as Familial adenomatous polyposis (FAP) and MUTYH associated polyposis (MAP) are known to predispose individuals to develop multiple polyps in their intestine, a condition known as polyposis. The presence of these polyps increases the risk of the individuals to develop premalignant and malignant tumours throughout their intestine. However, for some patients with a clinical diagnosis of polyposis, the current NHS genetic tests do not identify the genetic cause for their condition. These patients are eligible to enter a research study, led by Dr Hannah West, which is aiming to identify new genetic causes for polyposis.

The research study has recruited over 300 patients. The latest findings from this study, published in *The American Journal of Human Genetics* (available here), identified a gene called *MBD4* to be altered in some patients with polyposis. This has led to the proposal of a new rare genetic syndrome, MANS, and expands the genetic tests that can be offered by NHS clinical services.

New Diagnosis: the impact of research

These research findings can be translated into the clinical setting to expand the genetic testing offered for patients with polyposis and multi-tumour phenotypes to include *MBD4*. This is important because the improved diagnosis helps establish improved management plans for the patients affected by the specific and unique features of this condition.

Members of the Inherited Tumour Syndrome research team presenting their research findings at The European Hereditary Tumour Group conference 2021. From left to right: Laura Butlin, Laura Thomas, Hannah West.
You can read more about other studies supported by the Wales Gene Park Research coordinator on the Health and Care Research Wales Website: https://healthandcareresearchwales.org/about/news/we-refused-think-nothing-could-be-done-mum-son-affected-rare-disease-highlights

The role of a Research Coordinator
A research coordinator has a very specialised and important role within a research group. They often sit at the centre of a study, working in collaboration with the main investigator to oversee the work. Their role is often associated with the delivery of clinical trials, although they have an equivalently important role for research studies that recruit patients, or work with patient derived samples.

“The role of a research coordinator is so rewarding. I get to work with many different people from sponsors to patients and research nurses to produce revolutionary research that ultimately helps people living with rare diseases. The work we produce is vital for these patients and I get to be a part of that.”
Laura Butlin Wales Gene Park research coordinator

Research coordinators contribute to the smooth running and delivery of successful studies. They are critical in the patient recruitment process, and they serve as the link between the patients and the main investigator, ensuring informed consent is obtained and patient information is stored safely and securely.

Working with the main investigator, a research coordinator will produce compliant consent forms, develop the protocol, collect clinical data, liaise with hospitals/sites and acts as the main contact point throughout the duration of the study. They also work closely with the sponsor team and governing bodies to protect the safety of the patients and to ensure the study is ethically compliant.

“What makes a successful study?
Successful research is built on comprehensive research plans, allowing a study to run smoothly and efficiently. The plan includes details about how participants in the study are recruited and retained. Putting together an effective plan requires in-depth knowledge and expertise.

To further her own professional development, Laura, the WGP research coordinator, became a reviewer for “The ORRCA project (Online Resource for Research in Clinical triAls)” - https://orrca.org.uk/. This project aims to create a free online searchable database to help those responsible for developing study plans to quickly identify relevant literature on effective recruitment and retention strategies. The review is ongoing, but the first paper has been published (available here).

“The research coordinator has supported the research from the start, working with me to develop my research idea into a full ethics application, identifying potential issues with ethics, samples, and recruitment, and determining ways to overcome these. She has identified individuals to recruit and worked with diverse teams to recruit these individuals so that the study is properly powered to achieve its aims.”
Dr Laura Thomas, Swansea University.
Case Study 4
Genomics Showcase

The Genomics Showcase event was developed to highlight advancements in the field of genomics and the potential to improve healthcare to the people of Wales, in line with the Welsh Government’s genomics for Precision Medicine Strategy.

Plans for the first Showcase, scheduled for May 2020, were halted by the Coronavirus Pandemic and the event was cancelled. As the pandemic continued, it became clear that it would not be possible to hold a physical event in the near future, so a decision was made to take the event on-line. The aim was to produce an event which mirrored the experience of attending in person as closely as possible. The on-line platform provider, vFairs seemed to be the closest match to our requirements and Genero Productions, a creative events agency, was chosen to help us deliver the event.

As interest in the event spread and people were approached to take part, it became clear that the Showcase was developing into a much larger event than anticipated. The Showcase grew as the planning proceeded and eventually two talk areas developed into six. It was clear that attendees would not be able to access all the information available to them in one go, so the content was made available for 30 days after the live event in May 2021, allowing people to listen to talks that they missed on the day, at their own convenience.
On the day there was a total of 54 talks spread over 6 stages and a Genomics Café held over the lunchtime. The Exhibition Halls housed 35 stands and the Interactive Zone hosted 7 stands offering audience participation. 29 academic posters were displayed. The posters were judged and the best 6 were invited to make oral presentations on the day.

Prizes were awarded to the best poster and the best oral presentation. Over 1200 people registered for the Showcase, joining on the day and/or accessing the online content after the live event.

Feedback.

In free-typing feedback boxes, the platform received praise for its ease of navigation and for how it looked:

“My first experience of a virtual event and it worked really well - almost like attending the real thing, but from the comfort of my own chair!”

“I thought it worked very well. I am not very techy but had no problem in viewing the timetables and deciding which talks I wanted to attend. It was very straightforward logging in and out during the day.”

The content of the talks and speakers was also positively received:

“This is a wonderful way to showcase Genomics and inform a wider audience about a complex topic delivered by expert communicators.”

“Excellent talks and speakers. Thanks for a very interesting day.”

For future events, there was an appetite for a virtual element to be incorporated:

“I hope that future events would be online too alongside actual bodily attending. For those unable to get there in person for whatever reason virtual is great.”

“Please allow online participants at future events.”

The first Genomics Showcase was undoubtedly a success, meeting all the set objectives. Registration targets were exceeded, and feedback of the event has been overwhelmingly positive. It has been decided to hold the Showcase bi-annually and the next event is scheduled for 2023.

The Genomics Showcase programme and the Genomics Showcase report can be found here.
Case Study 5

Tremolo Podcast by Lisa Parry

A podcast drama for post-16 students and the public exploring the impact of a diagnosis of early-onset familial Alzheimer’s Disease (eFAD).

As genetics/genomics becomes increasingly vital in mainstream medicine/healthcare, enhanced public engagement and understanding is key. Creative approaches are increasingly recognised as stimulating, effective ways of communicating with the public about science. In education, this can help enhance learning and explore attitudes.

Tremolo is an exciting new collaborative project with Illumine Theatre and Theatre Genedlaethol Cymru. The project produced:
• A creative, thought-provoking 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. The bilingual podcast is free and aimed at young audiences aged 16+ throughout Wales, but available to everyone
• An accompanying bilingual educational resource pack aimed at year 12/13 students
• Tremolo was launched in March 2022 and is available to listen and download via platforms including Spotify, Apple Podcasts and AM.

Background

Wales Gene Park’s strong public engagement track-record has been combined with Illumine Theatre’s skills and experience to tell a powerful story. Discussions with schools informed the storyline and eFAD was chosen, bridging a gap between a subject often associated with an older demographic and younger people. Genetics specialists from the All-Wales Medical Genomics Service worked with playwright Lisa Parry to ensure the storyline’s scientific accuracy, whilst Theatr Genedlaethol provided creative input and funding to produce a Welsh/English bilingual drama. The project also received external funding from the Genetics Society and the Adelphi Genetics Forum public engagement award schemes.
Storyline

Tremolo is presented through the eyes of Harri, a caring teenage boy. Like many youngsters who have just completed their final A-Level exams, Harri is excited about the future; inter-railing with his friend and then off to Uni to follow his dream of becoming a neurosurgeon. Suddenly, his world is turned upside down when his loving Mam is diagnosed with eFAD. Due to genetics, there’s a 50% chance that he and his younger sister Gwenllian may have it too. As the play develops, we learn about the early signs of the condition. Through Harri’s eyes we also learn about the effect of the condition on family relationships, financial pressures, daily life and dreams.

As the name suggests, music has been an integral part of this production from the outset. Harri’s younger sister Gwenllian loves playing the harp and she spends hours practising her ‘tremolo’, and live harp music - composed and performed by internationally acclaimed Welsh harpist Eira Lynn Jones - was used to bring the story and emotions to life in this powerful production.

Educational Pack

The drama is also accompanied by an education pack for schools & colleges aimed at year 12/13 students. The pack contains scientific content (including an Information Toolkit, careers support, scientific activities, points for reflection and a Help and Support section) and creative content (including a synopsis of the drama, interviews with the creative team and creative activities). WGP is utilising its well-established national Schools’ Genetics Networks and extensive educational links to publicise the project and teachers/educators can get a copy of the pack by emailing walesgenepark@cardiff.ac.uk

Feedback

In its first month of release, Tremolo has been well received with very positive feedback from listeners. As well as a planned engagement event, the project will be evaluated over the coming months with findings disseminated to stakeholders.

“Families throughout our communities have to make decisions such as those taken by my main character, teenager ‘Harri’, and his family, and we rarely hear about it. I really hope Tremolo helps families considering genetic testing options and makes understanding these options more accessible to the general public.”

Lisa Parry, Playwright

“I have really enjoyed being in Harri’s world, I’ve learnt a lot from it. We don’t tend to think about serious conditions like early onset Alzheimer’s and how it affects families, until we experience it ourselves. I hope that hearing Lisa Parry’s harrowing script through Harri’s voice will raise awareness of the condition, spark discussions amongst young people and remind us what’s really important in our busy lives.”

Gareth Elis, Actor (Harri)

“With the new curriculum in Wales it’s a great chance for the expressive arts to connect with science and technology, and teachers can take advantage of a bespoke bilingual and free educational resource created by Theatr Gen and Wales Gene Park to support this podcast drama in the classroom.”

Rhian Davies, Executive Producer Theatr Genedlaethol Cymru

Tremolo is an Illumine Theatre production in partnership with Wales Gene Park and Theatr Genedlaethol Cymru, supported by the Genetics Society and the Adelphi Genetics Forum. Starring Gareth Elis. Written by Lisa Parry, directed by Zoë Waterman, harp music composed and played by Eira Lynn Jones, sound design and editing by Rhys Young, assistant director Branwen Davies, additional music by Yws Gwynedd. Tremolo was recorded at Hoot Studios, Cardiff.
Looking Forward

Supporting genomic research in Wales

Wales Gene Park activities are founded on the bedrock of collaboration. The resources needed to enable and support genomic research are expensive and are being updated at pace. The agile nature of Wales Gene Park, combined with the skill sets of the individuals involved, positions this Health and Care Research Wales Infrastructure support group to respond quickly to support researcher needs.

Wales Gene Park was established with support from the Welsh Government, the Department of Trade and Industry (DTI) and the Knowledge Exploitation Fund in 2002. We will soon be marking our 20th Birthday. Not only does this landmark allow us to reflect on what has been achieved, but it prompts a deeper review of what will be needed going forward.

There are several key enablers that Wales Gene Park can contribute to, and focusing on, as we move forward.

**Big data, IT, and expertise:** Handling genomic data at scale demands the right technology, workforce, and governance. Through the Sêr Cymru IT, the Wales Cancer TRE Project, and our established partnership with Genomics Partnership Wales, Wales Gene Park will continue to inform Wales’ use of genomic big data for the benefit of the Welsh people.

**Data integration:** Coordination and collaboration are key to the access, sharing, and linkage of medical data for healthcare research benefit. Wales Gene Park will continue to bring partners together and provide the direction and leadership needed to unlock the research and development potential of genomic data entrusted in academia and the NHS for the Welsh people.
Laboratory and Data Analysis Expertise: Wales Gene Park has always strived to attract and retain 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 both Wales Cancer Research Centre and the Experimental Cancer Medicine Centre in Cardiff to help them build their data scientist workforce. We also continue to fund staff embedded within the AWMGS and the SAIL databank.

Spatial Transcriptomics: Wales Gene Park were co-investigators for a successful funding bid that has allowed the purchase of a GeoMx® Digital Spatial Profiling platform in Cardiff. This piece of equipment is a novel cutting-edge platform which allows researchers to obtain a spatial profile of gene expression, either at the mRNA or protein levels, in tissue specimens. This means researchers can see in a tissue section which genes are expressed in which cells. This provides new insight for discovery and translational research looking at how cells interact with each other, how the genome activity varies across tissues, and how these differences can change in response to therapy. Wales Gene Park are developing the expertise needed to allow researchers across Wales to access this technology.

Outreach and Engagement Expertise: Wales Gene Park will continue the delivery of an extensive portfolio of events to promote health professional and public education and engagement. We will 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.

Cardiff Edge: Wales Gene Park are working closely with our Genomics Partnership Wales colleagues to implement and fulfil plans to see the establishment of a vibrant ecosystem of research, innovation and NHS Service Development at a new facility, North of Cardiff, at the Cardiff Edge site. Welsh Government are looking to establish a Centre of Excellence in Precision Medicine as a priority for Wales, and have invested in plans to see all partners from across the Genomics Partnership Wales, including All Wales Medical Genomics Service, Wales and Pathogen Genomics Unit (PenGU) and Wales Gene Park, to relocate to the new Cardiff Edge site next spring, as the first phase of these bigger ambitions.
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

As a Health and Care Research Wales funded group, Wales Gene Park comprises a team of genomic technologists, bioinformaticians and education and engagement practitioners, that constitute an essential partner within Genomics Partnership Wales, helping realise the ambitions of Welsh Government.

Wales Gene Park focuses on the provision of expertise and infrastructure needed to support a wide range of research and development projects in health priority areas throughout Wales.

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 29 publications within the reporting period, advancing scientific knowledge in a range of scientific fields. One such project, described in case study 3, highlights a route by which research can inform clinical diagnosis.

We are continuing to expand the portfolio of technologies supported by Wales Gene Park, which now includes the cutting-edge technique of examining spatial transcriptomics. The Wales Gene Park team are, however, small and demand for services, particularly for data analysis and bioinformatics, outstrips the capacity available.

The Education and Engagement team have again delivered a large programme of events for a range of audiences and enabled bidirectional connections to be made between researchers and the public or patients. This year, the events have predominantly been delivered online, although in person events have also been reintroduced to the programme. The team play critical roles delivering the Genomics Partnership Wales programme, and through our endeavours the Welsh Public have opportunities to be informed about the advances of genomic technologies in health care.

The benefits of investments in Wales’ digital infrastructure for genomics research are being seen. Building on the recently purchased Sêr Cymru IT infrastructure, Wales Gene Park have led a consortium of partners to improve Trusted Research Environment capabilities in Wales for cancer and rare genetic conditions. Our focus is on unlocking the potential of healthcare genomics data for translational research in a way that is safe, secure, and trustworthy.
# Glossary

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<td>3G</td>
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### A

| AML  | Acute Myeloid Leukaemia |
| AWMGS | All Wales Medical Genomics Services |

### C

| COVID-19 | Coronavirus Disease |
| CPD       | Continuing Professional Development |
| CRISPR    | Clustered Regularly Interspersed Short Palindromic Repeats |

### D

| DNA          | Deoxyribonucleic Acid |
| DTI          | Department of Trade and Industry |

### E

| ECMC         | Experimental Cancer Medicines Centre |
| eFAD         | Early-onset Familial Alzheimer’s Disease |
| ES           | Embryonic Stem |
| ExECT        | A Natural Language Processing Application used at SAIL |

### F

| FAP          | Familial Adenomatous Polyposis |

### G

| GB           | Great Britain |
| GPW          | Genomics Partnership Wales |

### H

| HDR-UK       | Health Data Research UK |

### I

| ILAW         | International League Against Epilepsy |
| iPSCs        | Induced Pluripotent Stem Cells |

### M

<p>| MAP          | MUTYH-Associated Polyposis |
| MDT          | Multidisciplinary Team |</p>
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<td>NDR</td>
<td>National Data Resource</td>
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<td>NGS</td>
<td>Next Generation Sequencing</td>
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<td>ORRCA</td>
<td>Online Resource for Research in Clinical trials</td>
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<td>PenGU</td>
<td>Public Health Wales Pathogens Genomics Unit</td>
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<td>PPI</td>
<td>Patient and Public Involvement</td>
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<td>QC</td>
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<td>RDIG</td>
<td>Rare Disease Implementation Group</td>
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<td>RNA</td>
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<td>Secure Anonymised Information Linkage</td>
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