Contents

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

Introduction

2 Welcome
3 Foreword
4 A Word from our Patient & Public Representatives
6 Strategic Objectives
7 Resources & Wales Gene Park Structure
9 Lay Summary

Work Packages

11 Work Package 1
Genome Editing & Transgenics
15 Work Package 2
Genomic Facility
21 Work Package 3
Education & Engagement

Case Studies

30 Applying Artificial Intelligence Techniques to Precision Medicine
31 Genomic Partnership Wales Patient and Public Sounding Board
33 Living with Genetic Conditions
35 Genomic Data Integration: Brief Summary
37 The 100,000 Genomes Project in Wales

www.walesgenepark.cardiff.ac.uk
Wales Gene Park is an infrastructure support group funded by Welsh Government through Health and Care Research Wales, hosted by Cardiff University.

This report presents activities from our 16th year of consecutive funding from Welsh Government. We are an integral part of Genomics Partnership Wales, an initiative that is delivering the Welsh Government’s Genomics for Precision Medicine Strategy.

Genomics Partnership Wales is bringing together partners to harness the potential of genomics to improve the health, wellbeing and prosperity of the people of Wales.

Wales Gene Park provides collaborative access to state-of-the-art technologies for researchers in Wales and provides innovative initiatives to educate and engage health professionals and the public in Wales and beyond.

Dr Karen Reed
Operations Manager,
Wales Gene Park
Foreword
Harnessing Genetics and Genomics to advance Research, Healthcare, Education and Innovation

When Peter Kille, Colin Lucas and I prepared the original bid to create the Wales Gene Park in 2001, the first draft sequence of the human genome was just being announced. Our bid anticipated the impact that genomics would have on the diagnosis and treatment of disease and it proposed the Wales Gene Park as a new organisation whose mission would be to ensure this opportunity was seized fully here in Wales. It has been a huge privilege to lead the Wales Gene Park over the intervening years as this hope has become a reality. Genomic science is now an essential cornerstone of biomedical research, while applications in healthcare have already gone well beyond what we could have imagined at that time. Stunning advances in technology have enabled this progress and the team at the Wales Gene Park has provided access to these technologies, supported researchers in their use and informed and involved professionals, patients and the wider population of Wales in the opportunities and challenges they bring. This annual report captures the value and impact of that work over in the year to April 2019. In the coming year as Dr Andrew Fry takes over as Director, ably assisted by Dr Karen Reed, Angela Burgess and the whole team, I have no doubt that Wales Gene Park will go from strength to strength and bring ever greater benefits to researchers, patients and the people of Wales. Pob lwc!

Professor Julian Sampson
Director, Wales Gene Park

This annual report captures the value and impact of the brand portfolio of work undertaken by the Wales Gene Park team.
I am a parent/carer with over three decades of experience of Autism, Learning Disabilities, Challenging Behaviour and Charitable Organisations. My youngest son has Tuberous Sclerosis Complex. I am a retired (early, I just look older) Developer Services Manager with Welsh Water, after 37 years in the Industry. I have been a patient representative on the Wales Gene Park Strategy Advisory Group for a couple of years now, it’s a way of giving something back to society and to an area I have an interest in.

By becoming involved I keep informed of what’s happening, within Wales and the other side of the Severn Bridge. I believe, that through Genetic Research great in roads have been made towards finding treatments and eventually curing many conditions.

WGP has an extensive programme of Educational and Public Engagement sessions that are arranged throughout Wales. These events attract interest across a wide range, professionals and public, giving opportunities for networking, sharing information and education.

I would encourage everyone to attend, they are very informative and cover varying topics, the sessions are targeted to specific age groups, professionals and general public.

Being a patient representative has enabled me to enhance my knowledge and understanding of Genomics and Genetics whilst ensuring that individuals and patients are at the forefront of diagnosis and treatment wherever possible. Genetic Research increases understanding of a condition, however, I believe, turning that understanding into treatments has to be the GOAL.

Perry James, Patient Representative

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It’s good to see that Wales Gene Park is positive and forward thinking in welcoming the patient onto the board. Indeed the patient is the expert in their own condition, so who better to directly interact with, to enhance the work of Wales Gene Park?

I am proud to be involved with WGP at this level, not only as a patient of a rare disease (Ataxia) but also as a patient group founder and advocate, so it’s a win, win situation. As I attend many conferences around the world it is very encouraging to hear the positive comments about WGP from around the globe and knowing that I have a little part to play in its presence.

Alan Thomas, Patient Representative
# Wales Gene Park

## 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 22 staff (17.1 whole 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 2018 - April 2019. Lines of formal communication are shown.

Budget breakdown 2018 - 2019 across Wales Gene Park activity
The Wales Gene Park annual budget employs 22 staff (17.1 whole time equivalent), who are supported by senior academics, experts from the NHS in Wales, managers and administrative staff from the host institution, Cardiff University.

**Key Partners**

**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
- Alexion Pharmaceuticals
- AstraZeneca
- Bayer
- BioVitas Capital Ltd
- Calithera Biosciences
- Chromatrap
- Gaia Technologies
- Genentech
- GW Pharmaceuticals
- Merck KGaA
- Myriad Genetics
- Nanoether Discovery Science Ltd
- New England Biolabs
- Novartis Pharmaceuticals

**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)

**Who’s Who**

**Strategic Advisory Group**
- Chair: Professor Colin Dayan, School of Medicine, Cardiff University
- Mr Ifan Evans, Deputy Director, Healthcare Innovation Welsh Government
- Dr Rob Orford, CSO Health, Welsh Government
- Professor Julian Hopkin CBE, Experimental Medicine, Swansea University
- Professor Malcolm Mason OBE, College of Biomedical and Life Sciences, Cardiff University
- Mr Alastair Kent OBE, Former Genetic Alliance and Rare Diseases UK Ambassador
- Dr Mick Hunter, CSO and Director of Development at Atopix and CEO at Orca Pharmaceuticals
- Dr Clive Morgan, Cardiff and Vale University Health Board
- Rachel Butler MBE / Sian Morgan, Head of Laboratory at the NHS All Wales Medical Genomics Service Laboratory

**Patient Representatives**
- Mr Perry James
- Mr Alan Thomas

**Executive Management Team**
- Professor Julian Sampson, Director
- Dr Karen Reed, Operations Manager
- Ms Sherrie Witts, Finance Manager
- Professor Mark Rees, Impact Lead
- Dr Kevin Ashelford, Bioinformatics Lead
- Ms Sarah Edkins, Genomic Laboratory Lead
- Professor Ros John, Genome Editing Lead
- Dr Ming Shen, Transgenics Lead
- Mrs Angela Burgess, Education and Engagement Lead
- Mrs Emma Hughes, Genetic Alliance Policy and Engagement Manager
Wales Gene Park is an infrastructure support group funded by Welsh Government through Health and Care Research Wales. We support research via three work packages and through the provision of technical support, research governance and patient recruitment activities associated with several genetics research studies registered on the Health and Care Research Wales Clinical Research Portfolio.

Work Package 1
Genome Editing and Transgenics
Making and using pre-clinical models of disease through genome editing and related methods

Work Package 2
Genomic Facility
Sequencing and analysing genomic information in collaboration with researchers

Work Package 3
Education and Engagement
Enhancing awareness and understanding of health-related genetic and genomic research for the public and health professionals

Lay Summary

43 Projects supported by Genomics Facility
325 TB Genomic Data
Samples Banked 417
Research Publications 40
Developing models of disease

Before new treatments can be trialed 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 research groups, in the areas of cancer, arthritis research, neurodevelopmental disorders, neurodegeneration, epigenetics and prenatal determinants of health later in life.

Pre-clinical trials

Wales Gene Park also undertakes pre-clinical trials for rare diseases in collaboration with several drug companies. This work uses genetically modified mouse models and human patient samples. Such trials have tested four new drug combinations for the treatment of kidney tumours, adding to the scientific knowledge and informing future clinical trials.

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 three UK Clinical Research Network (UKCRN) / Health and Care Research Wales Clinical Research Portfolio projects in rare diseases. Through banking samples from patients with cancer or rare diseases, we have also 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 43 research projects, 33% of projects were for cancer biology and/or rare disease-related research, 67% were for other common diseases including arthritis, diabetes 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 instigated 43 new research projects this year and provided access to computing capacity to a further 17 researchers. Wales Gene Park is now custodian to over 325 terabytes of sequencing data held on behalf of researchers across Wales.

Raising awareness

Genomics is fast becoming part of our health care system. The Wales Gene Park Education and Engagement team run many events to raise awareness and understanding of these advances. This year, through 74 events organised by Wales Gene Park, we have reached:

- 1078 Health Professionals: 27 events
- 1960 Schools/Colleges students/teachers: 5 events
- 2329 Public/Patients: 42 events

Additionally, Wales Gene Park’s interactive stand has been exhibited at 17 external events for the public, schools, professionals, patients and families.

Supporting Policy

Working with patients with rare disease

The Wales Gene Park Education and Engagement team oversee the Welsh Rare Disease Patient Network, ensuring patients are informed of, and effectively represented in, development of appropriate Welsh Government health and social care policies.

The Genomics for Precision Medicine strategy was officially launched by the Welsh Government in July 2017 setting out the plan for strengthening genomic medicine in Wales. Wales Gene Park contributed to the development and implementation of this strategy.
Work Package 1: Genome Editing & Transgenics

Purpose

Transgenics (making changes to the genetic material by introducing new DNA sequences) and genome editing (making direct changes to the genetic material) enable the generation of new pre-clinical models of disease. Wales Gene Park supports research by using these technologies for the production and use of pre-clinical models. While these pre-clinical models can take several forms, at Wales Gene Park we have expertise using genetically modified mouse models (GEMM’s), and 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.

The outputs from this work package span mechanistic to pre-clinical therapeutic research. The models generated form the basis for many external research funding applications. This work also generates evidence that informs the development of clinical trials in patients.

Core Activities

- Utilising CRISPR (Clustered Regularly Interspersed Short Palindromic Repeats) genome editing technology to generate new models of disease for research
- Growing human iPS cells
- Embryonic Stem (ES) cell derivation from genetically modified mouse models
- Consultation for researchers requiring support in ES cell and genome editing techniques
- Use of GM models for research into disease mechanisms and pre-clinical trials
Supporting research across Wales and beyond
“Testing new drug combinations to assess new treatment options for kidney tumours in TSC disease.”

Dr Ming Shen
Division of Cancer and Genetics, Cardiff
Progress
This year, in Wales Gene Park, we have expanded our portfolio of skills to include the use of human induced pluripotent stem cells (human iPSCs) technology for research. This follows a strategic shift and expansion of genome editing activity to better meet the current needs of the research community.

Working with Prof Nick Allen, Cardiff School of Biosciences, Prof Ros John, Wales Gene Park Genome Editing lead and Mrs B Allen, Genome Editing Research Manager, have established genome editing (using CRISPR technology) of human iPSCs as a new core Wales Gene Park activity. This work includes the culturing, CRISPR editing, characterisation and cryopreservation of novel edited cell lines.

Forty new lines of genetically modified human iPS cells have been established, targeting 4 genes implicated in neurodegenerative disorders. This includes the targeting of multiple genes at the same time within the same iPSC line, in order to investigate inter-dependency of gene functions in neurodegenerative diseases, thereby mirroring the complexity of these disease in a relevant pre-clinical model.

In addition to this new line of work, Wales Gene Park genome editing facility has also supported research groups with the previously generated transgenic mouse lines. This includes cryopreservation of embryos, (1 group), training in cryopreservation techniques (1 group), experimental embryo transfer for behavioural analysis (1 group), providing tissue for research (4 groups, 2 Cardiff University, 1 Swansea University, 1 Switzerland), training in surgical techniques (1 group).

Dr Ming Shen leads on pre-clinical trials for rare diseases and cancer using GEMMs for TSC-associated kidney tumours.

TSC (Tuberous Sclerosis Complex) is a rare disease that causes the growth of benign tumours in different organs of the body, including the brain, lungs, heart, kidneys, skin and eyes. Dr Shen’s research team investigate the role of several genes in tumour development in the kidney by conditionally deleting genes in the kidneys in models that are pre-disposed to develop kidney tumours. Also, in collaboration with several drug companies, they undertake pre-clinical trials involving new drugs, re-profiling of established drugs and testing new drug combinations that offer the prospect of improved treatment options.

Rapamycin is a drug that is currently used as a treatment for TSC and works by inhibiting one of the cellular pathway’s tumour cells rely on for growth. Dr Shen’s team have used GEMMs to test 4 different drug combinations that would target other cellular pathways needed for growth, including glycolysis and glutaminolysis which are pathways cells use for energy production. The latest findings have shown that while duel inhibition of glycolysis and glutaminolysis is effective at reducing tumour growth, there are notable side effects and this treatment wasn’t as effective as rapamycin treatment. These findings have important implications for developing new treatment options. This work has been published in Neoplasia this year.

Outcomes
• Advising on CRISPR guide design and techniques. (3 groups – 2 Cardiff University, 1 Bristol University GW4 collaboration)
• 4 publications adding to scientific knowledge
Work Package 2: Genomic Facility

Purpose
The Genomic Facility provides access to competitive and flexible Next Generation Sequencing (NGS) for researchers in Wales together with expert bioinformatics support.

By providing these technologies, Wales Gene Park improves the quality and quantity of genomic research that is undertaken in Wales, and ensures that a greater proportion of externally awarded research funding is spent in Wales. The Genomics Facility also supports innovation by contributing to collaborations with the Pharma and Biotech sectors.

Core Activities
- NGS of 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
- Contribution to healthcare professional education
Flexible competitively priced NGS for medical genetics and genomics research
“Working with WGP has allowed us to develop protocols for a novel application of NGS technology, furthering our research into DNA damage in cells. Measuring the induction and repair of DNA damage is fundamental to understanding the causes of cancer.”

Professor Simon Reed
Division of Cancer
and Genetics, Cardiff
Progress
Wales Gene Park supports research projects involving genomic analyses through several routes; i) the provision of NGS, ii) the provision of bioinformatics analysis, iii) the provision of computing capacity and iv) the provision of secure, robust data storage.

During 2018 - 2019, the team produced and provided NGS data for 43 research projects, from 27 different research groups, generating nearly 3TB (terabytes) of sequencing data. 33% of projects were for cancer biology and/or rare disease related research, while common diseases including arthritis, diabetes and Alzheimer’s research accounted for 67% of projects.

The Wales Gene Park bioinformatics team have provided analysis and/or support for 97 projects during the 2018-2019 reporting period. 35 projects (36%) required an in-depth bespoke analysis, 15 (16%) were analysed using standard workflows, 5 (5%) involved providing advice and training to researchers, 5 (5%) for development project work and 37 (38%) involved quality control analysis of sequencing data before the data was sent to researchers to perform their own detailed analyses. 54% of projects were for research involving common diseases including cancer, arthritis, diabetes and Alzheimer’s. 14% of projects involved rare disease research and 14% were concerning infection and immunity. Of the 43 new research projects instigated in the last year, all have involved analysis on data generated in-house by the Wales Gene Park Genomics Laboratory.

Novel analysis methods employed by the bioinformatics team over the last year have included applications of machine learning in biomarker discovery, identification of novel microRNAs, the extraction and identification of T-cell receptor sequences from RNAseq data and single cell RNAseq analysis.

The team also successfully generated and analysed data from FFPE (Formalin-Fixed Paraffin-Embedded) tissue samples demonstrating new opportunities for the both DNA based (exome-seq) and RNA based (RNAseq) workflows to be applied to this expansive archive of patient samples.

Wales Gene Park has provided access to our supercomputing capacity to a further 17 researchers in 2018-2019, with a total of 34 researchers now utilising this specialist computing resource. Wales Gene Park is now custodian to over 325 terabytes of sequencing data held on behalf of researchers across Wales and has provided 304 terabytes of storage to other Welsh research groups.

Outcomes
• NGS provision via collaborative partnerships for 43 research projects from 27 different research groups
• Provided specialist supercomputing infrastructure to 34 researchers
• In collaboration with the Human Gene Mutation Database (HGMD) we have developed a new machine learning algorithm to better automate curation of databases relied on for genetic diagnosis (See Page 30 for more details)
Welsh Government have supported the purchase of the state-of-the-art Illumina NovaSeq 6000 sequencing platform, housed within the All Wales Medical Genomics Service - AWMGS, for diagnostic and research sequencing provision.

Wales Gene Park also continues to contribute to the broader development of IT provision for genomic medicine in Wales, through the Genomic Partnership Wales IT implementation group.

Wales Gene Park are liaising with AWMGS, host of the NovaSeq platform, to develop the strategy for sharing high throughput sequencing resources to support the research communities in Wales.

Working with NHS

Wales Gene Park works closely with All Wales Medical Genomics Service (AWMGS), providing resources and expertise to support the implementation of new high-performance compute and storage for diagnostic genetic service provision in the NHS.

New computer hardware has been installed within Cardiff University’s ARCCA (Advanced Research Computing at Cardiff) facility and is currently being configured and tested with the view of being ready for routine use in Autumn 2019.
The Nova Seq 6000 system provides high throughput state-of-the-art flexible NGS sequencing to support clinical and research applications in Wales.
Work Package 3: Education & Engagement

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

Core Activities
- Education and engagement events via public and schools programmes
- 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
Enhancing awareness and understanding of genetics and genomics in Wales
“Through sharing our personal experiences of family life and living with a rare genetic disorder, I have been empowered. Sharing my experiences with students and others who will provide care for families like mine is so important; knowledge is key to improve future outcomes for my son and others who live with a genetic disorder.”

Participant
Wales Gene Park Event
Progress

The Genomics for Precision Medicine Strategy sets out the Welsh Government’s plan to create a sustainable, internationally-competitive environment for genetics and genomics to improve health and healthcare provision for the people of Wales. Wales Gene Park is leading the way in educating and engaging health professionals, young people, patients and the public in genomics to ensure Wales is at the forefront of delivering health and care services and informing policy and research developments.

Public

A range of events was held for public and patient groups.

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

Over 220 members of the public attended events in Cardiff and Wrexham, enjoying talks on DNA, genetics, genomics and associated topics and interactive stands.

Community Group talks

Thirty-four DNA and genetics-related talks were delivered to community-based organisations throughout Wales including The Rotary Club, Women’s Institute groups, NHS Retirement Fellowships, Probus Clubs, University of the Third Age and library groups.

Public lectures

Attended by over 340 people, events included ‘The Future of Cancer Treatment: personalised medicine and how it is revolutionising patient care’; ‘The Welsh plan to transform patient care with genomics: panel of speakers from Genomics Partnership Wales and The 100,000 Genomes Project in Wales’; a screening of the film Wonder to mark Rare Disease Day 2019. The event included a personal account from a Welsh parent with a son affected by Moebius Syndrome, a rare condition causing facial paralysis.

Patients

Rare Disease Patient Network Annual meeting

Wales Gene Park held its annual Rare Disease Patient Network meeting in October 2018, bringing together patients, families and professionals to network and share information. Participants in the Health and Care Research Wales funded SIGNAL project, including a number of families who had received a diagnosis through the project, attended. The event included presentations from Genomics Partnership Wales and from patients and carers affected by rare conditions, as well as a workshop in collaboration with the School of Social Care Research in Swansea University to prioritise research into rare diseases.

Rare Resources Workshop

In January 2019, Wales Gene Park and Genetic Alliance UK invited families affected by rare, genetic or undiagnosed conditions to take part in a Rare Resources focus group in Cardiff: “Design the Toolkit”. Parents and carers had the opportunity to share their experiences to help design information resources for families across Wales.

Rare Disease Day Parliamentary Reception

On Thursday 14 February, Genetic Alliance UK held the first of its 2019 parliamentary receptions in the Senedd, Cardiff, hosted by Darren Millar AM. The event was well attended by stakeholders from across the rare disease community. Attendees were provided with an update on progress made delivering the Welsh Rare Disease Implementation Plan from Dr Graham Shortland. Dr Andrew Fry spoke at the event about the challenges and opportunities of genomics in Wales for rare disease families. Attendees also heard from three patient and parent speakers. Fern Cockrell and her mother, Sally-Anne Ashdown gave their perspectives of living with chronic inherited pancreatitis – a condition that means Fern is in constant pain.

Fern’s story also received a lot of coverage in the Welsh media - notably:

- BBC Radio Wales.
- BBC Wales Online.
- Wales Online.

Latifah Charles provided a parent’s perspective of sickle cell disease, raising awareness of the condition and its effects, and highlighting the need for improved services and care.

Another parent representative also had the opportunity to raise awareness of her son’s experiences of the rare condition tuberous sclerosis complex on BBC Post Prynhawn (a Welsh language drive time news programme).

Genetic Alliance UK’s new ‘Illuminating the Rare Reality’ report was launched to coincide with Rare Disease Day 2019. This report brings to light the difficulties people living with a rare disease face in accessing the care and treatment they need, and underpins the call for a refresh and review of the UK Strategy for Rare Diseases. To highlight the ‘Rare Reality’, Genetic Alliance UK also created a microsite where almost 1500 people shared their #RareReality by adding a star to the website, to help shine a light on the realities of living with a rare condition.

Sickle-cell and thalassaemia patient, carer, family and education day

A Wales patient, carer and family and education day was held for patients and families affected by sickle cell and thalassaemia. The event focused on improving services in Wales.

Rare Inherited Eye Disease event

In March, Wales Gene Park supported the organisation of an event that brought together patients, carers, patient organisations, health professionals, researchers and policy makers to share information and provide networking opportunities for patients and families affected by rare eye conditions. Held in collaboration with the School of Optometry at Cardiff University, topics included gene therapy, genetic testing, support and rehabilitation as well as personal stories from patients and carers living with rare eye conditions.

“Promoted interest in the subject. Would love to follow up”

Attendee, 3G
Health Professionals

Living with Genetic Conditions (see case study for further details)

Sessions which focus on personal experiences from patient and family members were delivered at the University of South Wales and Cardiff Metropolitan University and attended by over 320 nursing, midwifery and Allied Health Professional students.

Spotlight on Genomics

The two-day event, held in Cardiff in June as part of the Wales Festival of Innovation 2018, brought together over 75 experts to present the latest advances in genome editing and next generation sequencing.

Tuberous Sclerosis Update Day

A specialist meeting was held in Cardiff to update experts on the advances in the management of tuberous sclerosis that are transforming patient care.

Clinical Genetics Revision course

Hosted by the All Wales Medical Genomics Service, the annual course was held for doctors training and specialising in this area of medicine.

Tuberous sclerosis study day

WGP facilitated an accredited study day for health, education and social care professionals which was aimed at supporting children and adults with tuberous sclerosis complex.

Schools and Colleges

A varied programme, aimed at supporting genetics and genomics secondary education and raising awareness of relevant careers.

Sixth Form Genetics Conferences

Over 2000 year 12 and 13 students from schools and colleges throughout Wales attended the conferences, which took place in Cardiff and Wrexham.

Students heard talks from expert speakers on cutting-edge topics, and stands at both events included hands-on activities and genetics-related careers information.

Teachers’ Genetics and Genomics Continuing Professional Development event

Thirty five secondary biology teachers attended a ‘Cutting-Edge Biology’ CPD event run in conjunction with Techniquest, Swansea University and STEM Learning. Attendees heard updates on genetics and genomics topics including stem cells, epigenetics and cancer genomics and bioinformatics.

Film screening

Over 130 year 8 and 9 pupils from schools across south Wales attended a special screening of Jurassic World: Fallen Kingdom hosted in collaboration with Into Film. The film was followed by a talk on genetic engineering and cloning from Dr Rhys Jones.

“...amazing. Very open about the challenges of having a child with a rare illness. The film was excellent for showing the impact on different family members and friends”

Attendee, Wonder screening and talk

This event is an invaluable source of information that pupils would not be able to access without your time and effort.

Attendee
Cardiff 6th Form Conference

www.walesgenepark.cardiff.ac.uk
Policy

Genomics for Precision Medicine Strategy

Wales Gene Park continues to support the implementation of Welsh Government’s Genomics for Precision Medicine Strategy. We have worked with Genomics Partnership Wales, supporting recruitment to its Patient and Public Sounding Board, ensuring developments are focused on maximising patient benefit. Activities to date have included an induction day, and a consultation led by the Genomics Partnership Wales Consent Implementation Group on consent models for genomic research studies.

Welsh Rare Disease Implementation Plan

Wales Gene Park’s role as third sector representative on the Rare Disease Implementation Group (the group with oversight for implementation of the Welsh plan) continues. Wales Gene Park supports work streams focused on empowering patients and families and rare disease research, strengthening the programme around patient and public involvement in research through development of the Patient Rare Disease Research Portal.

Research

Rare Disease Patient Research Portal

The portal has been developed following feedback from the Rare Disease Patient Network meeting in 2017 and will be usability tested by patients and families in June ahead of its launch in autumn. It will contain Health and Care Research Wales Portfolio studies across genomics and rare diseases as well as other relevant non-portfolio studies.

SiGNAL Project

Participants and family members from the SiGNAL research project attended WGP’s annual Rare Disease Patient Network meeting in October to find out more about the results of the study and receive further information and support.

Priority setting for social care research for rare diseases

At the Rare Disease Patient Network Annual Meeting, Wales Gene Park ran a collaborative workshop with the School of Social Care Research at Swansea University and event attendees. The workshop set priorities for social care research for rare disease patients and families. Findings from the report will be utilised to identify new funding opportunities for rare disease and social care projects.

“WOW! Brilliant talk, so informative and ‘down to earth’. Again would love to spend a day with her (and my 6th form)”

Attendee
Teachers’s CPD Event
Outcomes

Wales Gene Park Education and Engagement team organised 74 different events between April 2018 and March 2019 in which 5367 people took part. Many more visited the Wales Gene Park’s interactive stand which has been exhibited at 17 external events. In total, we have reached:

- **Health Professionals**
  27 events & 1078 attendees
- **Schools/ Colleges**
  5 events & 1960 attendees
- **Public/ Patients**
  42 events & 2329 attendees

Example events that the interactive stand visited include the Youth Cymru STEM event for British Science Week, Technovate (part of the Welsh Festival of Innovation 2018) and the Cardiff Science Festival.

Wales Gene Park has also continued to increase its genetic and genomics networks for teachers, professionals, patients and the public.

As part of Genomics Partnership Wales, Wales Gene Park’s Education and Engagement team has also delivered a programme of activities throughout Wales to raise awareness of The Welsh Government’s Genomics for Precision Medicine Strategy. The programme, which has included genomics roadshows in each Health Board in Wales, stands and talks, has helped inform the Welsh healthcare workforce of advances in genomic medicine.

"Will definitely incorporate information into teaching"

Attendee, Teachers’s CPD Event
“Great day, fantastic speakers. Would love to attend to similar meeting again”

Attendee, TS Update Day 2018
Case Study
Applying artificial intelligence techniques to precision medicine

The Human Gene Mutation Database (HGMD) which is hosted by the Institute of Medical Genetics at Cardiff University and Wales Gene Park are working together to apply Natural Language Processing (NLP), a form of computer Artificial Intelligence to screen scientific papers to identify reports of human disease-causing mutations for the database.

HGMD is a major database used throughout the world both by healthcare professionals and researchers, providing up-to-date information on the mutations that cause or influence inherited disease. Keeping HGMD up to date in the genomic era is a huge challenge, with tens of thousands of potentially relevant scientific papers being published each year.

Drs Kevin Ashelford and Anna Price from Wales Gene Park have worked with Dr Matt Mort, Senior Software Developer and Analyst with the HGMD, to create software that automatically identifies research papers containing this information.

In addition to the advantages implemented in HGMD, the software promises to have wider applications in healthcare and research that are also being explored. Modern healthcare and research require integration of diverse sources of information. Much of a patient’s medical information such as the results of DNA sequencing, blood test results, images generated by medical scanners, prescribing and GP records is already curated in standardised ways on computers and can be relatively easily linked together.

But other information in letters, referral emails, medical reports, or indeed the research papers that HGMD are interested in, can be far more difficult to access by computer and requires new approaches such as NPL to be integrated.
Case Study
Genomic Partnership Wales
Patient and Public Sounding Board

The Welsh Government’s Genomics for Precision Medicine Strategy sets out an ambitious plan “to create a sustainable, internationally-competitive environment for genetics and genomics to improve health and healthcare provision for the people of Wales.”

Wales Gene Park is an integral member of the “Partneriaeth Genomeg Cymru – Genomics Partnership Wales” which was established in 2018 to ensure a united approach to genomics in Wales and represents several disciplines coming together to deliver a programme of work that will realise the commitment outlined in the Strategy.

The first of five Strategy themes is Co-Production, which describes Genomic Partnership Wales’s commitment “to work in an open and transparent manner with patients and the public in Wales, using their collective experiences to shape and add value to the work of the Genomics Partnership and future genomics services in Wales”.

Three high-level objectives were agreed:
1. Genomics Partnership Wales will work with patients and the public to improve the public understanding of genomics, ensuring that it becomes an accessible area of healthcare for the citizens of Wales
2. Genomics Partnership Wales will work with patients and the public to co-produce an open, transparent and publically agreed approach to the sharing of genomic and precision medicine data for service development, clinical care and research
3. Genomics Partnership Wales will use the experiences of patients and the public to improve the patient experience throughout the patient pathway, from the first interaction to the last

The aim of the Patient and Public Sounding board is to establish a meaningful approach to Patient and Public Involvement, which differentiates between Involvement and Engagement, ensuring the patient and public voice shapes our work and informs the next steps across the breadth of the Genomics Strategy implementation programme.

Ten members representing a range of patient experiences and diversity in terms of age, gender and Welsh region (including bilingual members), were appointed to the Sounding Board over the period Jan-March 2019. A two-day induction event and consultation followed. An exciting programme of work has been set for the next year, with quarterly consultations scheduled on diverse topics, including the review of consent models associated with research studies involving genetic testing. Additionally, two members from the Sounding Board have been appointed to Genomic Partnership Wales’s Governance and Programme Boards. This will ensure an equal voice for patients and the public in the work of Genomic Partnership Wales, at the heart of the decision making.
“I have been committed to establishing a strong patient and public involvement strategy to shape the work of Genomics Partnership Wales, since the day I was appointed to my role as Head of Programme.

A great strength of this Sounding Board is its diversity; collectively members represent a range of patient experiences - from rare diseases and cancer, to genetic testing during pregnancy, and they also represent a geographical spread across Wales. Some are experienced in Patient and Public Involvement and others are new to it. These are some of the reasons that I am excited to see this co-production evolve, building on strong foundations set by this Sounding Board to support the work of the Genomics Partnership in Wales, and I’m grateful to the Wales Gene Park for their support.”

Dr Catrin Middleton
Head of Programme - Genomics for Precision Medicine Strategy
Case Study
Living with Genetic Conditions
Promoting knowledge and understanding of rare and genetic conditions through personal narratives

The power of personal stories is widely recognised, being increasingly used in health to capture the individual’s voice and experiences and promote patient-centred care.

Established in 2008, Wales Gene Park’s ‘Living with Genetic Conditions’ informs others about genetic conditions and, through personal experiences, promotes understanding of the impact they have on everyday lives and on healthcare practice. Living with Genetic Conditions is aimed at educating others through the personal experiences of those affected by rare and genetic conditions.

**Approach**
Sessions begin with a genetics expert, usually a Genetic Counsellor or another specialist, giving a talk covering some basic genetics, information about the All Wales Medical Genomics Service and details about the genetic condition on which the session will focus. This is followed by a talk from someone affected by a genetic condition – such as an individual with the condition, a family member, or carer.

Their personal narrative gives an insight into the impact the condition can have on an individual or family, providing a powerful account of how it can affect them on a daily basis.

For example, speakers often describe their diagnosis, the treatment and management of the condition and how it can affect them on a social or psychological level. Participants choose what they would like to include when speaking about their personal experiences, and they can also use the opportunity to signpost to resources and support groups.

Educationally sessions help link theory and practice and highlight the relevance of genetics to healthcare. A question and answer session with the speakers usually elicits a range of interesting questions from the audience. Sessions last no longer than an hour, making them suitable for lessons, lectures or public talks.

Examples of genetic conditions that have been the focus of sessions include cystic fibrosis, tuberous sclerosis complex, sickle cell disease, Fragile-X syndrome, Lynch syndrome and Huntington’s disease.

"Someone with a genetic disorder giving us lectures makes me understand it well”
Nursing student
“Through sharing our personal experiences of family life and living with a rare genetic disorder, I have been empowered…..sharing my experiences with students and others who will provide care for families like mine is so important; knowledge is key to improve future outcomes for my son and others who live with a genetic disorder”

Speaker - Family Member

Feedback and evaluation
Originally intended for year 12 and 13 school and college students, its scope has been extended to include higher education and the public due to its success.

It is now delivered to undergraduate nursing, midwifery and Allied Health Professional students at Welsh universities including Cardiff, University of South Wales, Swansea and Cardiff Metropolitan with expansion to other Welsh HEIs planned. Sessions are received very positively by both students and educators.

Evaluations have shown the vast majority of students report that they feel more informed about genetics and want to know more about the subject as a result. Feedback from speakers indicates that this is also a valued opportunity for genetic counsellors to enhance their teaching experience and highlight the role of genetics services.

For individuals and family members, it is an opportunity to have a voice and improve knowledge and understanding amongst future health professionals and others of what it is like to live with a genetic condition.

Living with Genetic Conditions has been well received by lay audiences as part of WGP’s programme of community talks and public lectures.

It has also been delivered to younger audiences including Summer School participants, and as a Young People’s Panel at WGP’s Sixth Form Genetics conferences.

Furthermore, these personal accounts have formed part of the annual Welsh Rare Disease Day Senedd reception, highlighting issues faced by the rare disease community to a range of stakeholders including Assembly Members, policy-makers, industry representatives, clinicians, academics and the public.

Added benefit
• Conference abstract submissions and poster presentations (including European Conference on Rare Disease and Orphan Products)

• An extended ‘Spotlight on Rare Diseases’ showcase for nursing undergraduate students at the University of South Wales

• A participant-focused event to share success with the patients, families and health professional involved and provide them with an opportunity to shape future sessions

• Condition-specific, follow-up events for patients and families e.g. Lynch syndrome Patient Day to take place in September 2019

• Bringing together members of the Rare Disease community in Wales

Summary
As well as raising awareness and understanding amongst audiences, Living with Genetic Conditions provides an important outlet for the voices of those affected by rare and genetic conditions.

If you are interested in finding out more about our Living with Genetic Conditions sessions, please get in touch: walesgenepark@cardiff.ac.uk

Undergraduate Healthcare Students through university teaching sessions

1836

Members of the public through lectures and community talks

232

School/College Students through summer schools and the 6th Form Genetics Conference

1549
Case Study
Genomic Data Integration: Brief Summary

The Genomic Data Integration (GeDI, known to its friends as ‘Jedi’) project is funded by the MRC as part of the 100,000 Genomes Project in Wales.

Aim
The GeDI project aims to scope issues pertaining to the integration of genomic data into data safe havens, particularly the SAIL (Secure Anonymised Information Linkage) Databank, and its linkage to routinely collected health and social care data for use in anonymous form for research.

Study plan
The main components of the study plan were:

• An assessment of the status of genomic data in data protection law; in particular, the General Data Protection Regulation (GDPR).

• A literature review and mini-case studies examining how genomic data are accessed and used with health data.

• A series of eight workshops to gain public views on the reuse of genomic data collected for research. Groups were sourced with the assistance of the Wales Gene Park.

• The development of a set of flexible control measures that can be applied to mitigate disclosure risks and maintain data utility.
Summary of findings

• To date, the majority of genomic data arise from research, but it is anticipated that in future there will be more genomic data in the routine health record.

• The literature review showed the majority of published studies using genomic data are based on an external data release model.

• Mini-case studies of data centres showed a trend moving towards the use of genomic data within data safe havens.

• Our work indicated public support for the use of genomic data with health data, providing data are handled properly – data safe havens being the preferred model.

• Whilst genomic data are often unique this should not be equated with identifiable as it depends on how data are managed.

• Taking into account the varying granularity of genomic data and other risk factors (such as rare conditions), a flexible risk-control-utility model is needed for the optimum use of genomic data with health data in a safe haven.

Further work

Wales Gene Park and SAIL will work together to document practical worked examples of pathfinder projects integrating genomic and health data into SAIL, including:

Swansea Neurology Biobank epilepsy genomic data
Welsh Cancer Research Centre and Wales Cancer Bank
National Centre for Mental Health
100,000 Genomes Project data on Welsh patients

Fuller information is available on request.
Case Study
The 100,000 Genomes Project in Wales

Wales 100,000 Genomes team reached their recruitment target on time....
The project has enabled access to diagnostic whole genome sequencing for NHS patients in Wales who have suspected but currently undiagnosed rare genetic disorders. In doing so it is transforming the NHS in Wales into a service that can apply the results of whole-genome sequencing in everyday healthcare. This is providing a quicker, more accurate diagnosis of genetic disorders and better personalised treatments to patients across Wales.

Results
Recruitment into the project is now closed. In total, 439 patients from 154 families took part in the Welsh arm of the project. All the participants had previously undergone extensive investigation without a diagnosis being found. Analysis of the data is still ongoing, but already diagnoses are being made and fed back to patients and their doctors. With data still being analysed, the number of families who finally receive the answer they’ve been waiting for will continue to rise.

Award Winning Team
The 100,000 Genomes Project Team was nominated for and won the 2019 MEDIC STAR award in the category of the best research team. The MEDIC STAR award is presented by Cardiff University every year to celebrate team and individual achievements in research, teaching and citizenship in the School of Medicine. In the picture below you can see Dr Iris Egner and Mr Rhys Vaughan receiving the award from Professor Gary F. Baxter, Pro Vice-Chancellor (on right) and Professor Simon Jones, Dean of Research in the School of Medicine (on left).
Achieving Impact

Wales Gene Park works to help Wales realise its ambitions for the impact of genomics in line with the Welsh Government’s 2017 Genomics for Precision Medicine Strategy.

As an infrastructure support group, Wales Gene Park strives to achieve the greatest possible reach and significance for the impact of its work by:

- Providing researchers in Wales from HEIs, NHS and commercial sectors with access to Next Generation Sequencing, bioinformatics and genome editing.
- Working with researchers to increase the quality and quantity of genomic research in Wales to grow the significance and reach of associated impact.
- Improving understanding in the NHS workforce and amongst patients and the wider public of the opportunities and challenges presented by genomic medicine.
- Working with NHS genetic and genomic services and clinical researchers to ensure efficient knowledge transfer from research to diagnostic provision for better and faster genetic and genomic tests.
- Facilitating the development of well-informed and effective policies for genomic medicine by Welsh Government and other organisations.
- Participation in working groups, taskforces and the provision of expert advice.
- Helping patients, their families and carers, and the wider public to have their views represented.

£3.1M

Grant Income Supported
20 Successful Grant Awards
Dr Hywel Williams (Cardiff University) has taken up the role as Wales Gene Park Impact Lead from Professor Mark Rees (Swansea University).

Dr Williams will work with the Operations Manager, Dr Karen Reed, who liaises with the wider Health and Care Research Wales infrastructure to identify areas for joint working to achieve the greatest reach and significance for research.
Key Achievements
2018-2019

Event Attendees

5,367

Our Education and Engagement Programme reached 5,367 people across 74 organised events by Wales Gene Park.

Network Members

1,234

We have expanded the Wales Gene Park Patient Network to 141 members, the Public Network to 647 members and the schools/teachers network to 446 members to facilitate public engagement and involvement in genetic and genomic research.

Grant income supported

£3,092,838
Genomics Facility

Sequencing

43
Projects

- Cancer & Rare Diseases: 32%
- Common Diseases: 68%

Bioinformatics

97
Projects

- Bespoke analysis: 35
- Standard analysis: 16
- Advice and training: 5
- QC and Packaging: 37

Supercomputing Infrastructure

34
Researchers

325Tb WGP NGS data

304Tb WGP storage used by others

2,838
Looking Forward
Supporting genomic research in Wales into the 2020s

As we put the final touches to this year’s annual report, we are awaiting the outcome of the funding application made to Health and Care Research Wales. In the application, we set out our vision for the next five years (2020-2025), building on our commitments to support the delivery of Welsh Government’s strategies, including the Genomics for Precision Medicine strategy and the Implementation Plan for Rare Diseases.

We will provide the technologies required by Welsh researchers into the next decade:

• Partnership working to ensure the provision of NGS and bioinformatic facilities and expertise to support the needs of health researchers in Wales, including research undertaken in partnership with the NHS and with the commercial sector

• Human iPSC derivation and genome editing to support researchers needs through the provision of appropriate pre-clinical models of disease to understand the causes of disease and to develop and test potential treatments

The Rare Disease Patient Research Portal, to be hosted on the Wales Gene Park website, has been developed to help patients and public find opportunities to become involved in research.
• Working with the experts in e-health research at the SAIL (Secure Anonymised Information Linkage) databank at Swansea University, and the Genetic Data Integration (GeDI) project team, to establish methods for interfacing genomic data with clinical and other e-health related data for research innovation and service development

• Delivery of an extensive portfolio of events to promote health professional and public education and engagement, and to facilitate the informed involvement and participation of patients and public in setting research priorities in health and social care and in research planning, funding, dissemination and translation

• Service and policy development work, working with Genomics Partnership Wales, the new Welsh Assembly Cross Party Group for rare, genetic and undiagnosed conditions (launching in 2019) and other partners to influence and develop policy areas which improve the lives of patients and their families

The next generation of leaders

The current Wales Gene Park Director, Prof Julian Sampson, plans to step down in September 2019, creating an opportunity to pass Wales Gene Park leadership to a new generation. The Directorship will be taken up by Dr Andrew Fry, a Clinical Senior Lecturer and Honorary NHS Consultant in Medical Genetics, working closely with proposed WGP Co-Directors, Dr Karen Reed (Operations Manager and Lead for Genomics in Research) and Mrs Angela Burgess (Lead for Education and Engagement). These changes align with the five-year vision of Wales Gene Park to play a leading role in delivering the Welsh Government’s objectives for research, innovation, and engagement to progress the mainstreaming of genomics for improved health and wealth generation.

www.walesgenepark.cardiff.ac.uk
Aknowledgements

Many senior academics, university management and NHS staff continue to support Wales Gene Park, giving their time, energy and enthusiasm. We thank them for their invaluable help. We further thank Mr Perry James and Mr Alan Thomas, our patient representatives, for their continued excellent support and advice.
Conclusion

The momentum in Wales to utilise genomics within healthcare and healthcare research is growing. Genomics underpins the development of precision medicine for quicker and more accurate diagnosis of disease and for stratification and personalisation of treatment. Wales Gene Park is an essential partner within Genomics Partnership Wales, helping realise the ambitions of Welsh Government, Higher Education Institutions, the NHS and industrial partners to make advances and to develop and deliver better quality health and care services in Wales.

Wales Gene Park comprises a team of genomic technologists, bioinformaticians, genomic medicine and education and engagement practitioners, who together are committed to support the delivery and implementation of the Welsh Government’s Genomics for Precision Medicine strategy and the Welsh Rare Disease Implementation Plan. As an infrastructure support group, we focus on the provision of expertise and infrastructure needed to support a wide range of research and development projects in health priority areas throughout Wales.

We are supporting researchers by expanding our portfolio of technologies to include cutting-edge techniques using patient derived tissues, including human induced pluripotent stem cells.

The continued development of appropriate models of disease, underpinning exploratory research in the pre-clinical setting, is essential to support the fundamental stages of the translational pipeline. Complementing this, Wales Gene Park ensures the provision of competitively priced, bespoke NGS and bioinformatics for researchers across Wales, seeing grant income into Wales is spent in Wales.

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. The Wales Gene Park team have played critical roles delivering the Genomics Partnership Wales health-care professional roadshows and supporting the development of the Genomics Partnership Wales Patient and Public sounding Board.
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<td>3G</td>
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<td>ARCCA</td>
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<td>CPD</td>
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<td>CRISPR</td>
<td>Clustered Regularly Interspersed Short Palindromic Repeats</td>
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<td>Embryonic Stem</td>
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