

## The Welsh genomics revolution gains momentum in 2019

Firstly, thank you all for a fantastic first year as Genomics Partnership Wales. I'm incredibly proud to be part of such a collaborative, hard working and committed group.

In September, we reached a significant milestone by surpassing our target contribution to Genomics England's 100,000 Genomes Project. Wales's effort not only enabled our patients to access cutting edge research and potential subsequent diagnoses, but also demonstrates our commitment to work collaboratively across the UK.

In November, the Pathogen Genomics Unit hit the headlines thanks to their swift, world-leading analysis of the flu virus. We also held our first public engagement event, Cardiff University's Public Understanding of Science in Health (PUSH) lecture series, with all partners united as GPW. The first of many I hope!

2018 was about building the strong foundations of our new partnership. 2019 will be about driving momentum, delivering on key aspects of the Genomics for Precision Medicine Strategy; Co-production with the public, Clinical & Laboratory Services, Research & Innovation, Workforce, and Strategic Partnerships, to ensure that we remain forward-thinking and that our services can grow and prosper in Wales.

We're currently recruiting for the first Patient and Public Sounding Board, where real-life experiences can shape our work. The future of genomics in Wales is looking bright, I'm sure you'll agree!



*Len Richards*

**Len Richards**  
Chief Executive  
Cardiff and Vale UHB  
Senior Responsible Officer  
GPW

## GPW In Focus

### Pathogen Genomics Unit world leader in identifying flu strains

Public Health Wales Pathogen Genomics Unit (PenGU) has been praised as a world leader in the global mapping of flu.

Reporting on the department's swift analysis of DNA sequences of the virus, BBC News highlighted the efforts of the unit in providing the latest data to scientists who are tracking the way flu spreads and evolves across the world.

Early identification of the H1N1 (pdm 2009) strain of flu virus will help scientists to better track changes in the virus and ultimately help to produce a more effective vaccine for future flu seasons.

Speaking to the BBC, Dr Tom Connor, bioinformatics lead at PenGU, confirmed that the majority of samples had come from Wales, sitting alongside Hong Kong and New Mexico as the key contributors of data.

He added, "the opportunity to have an impact on a worldwide scale was 'phenomenal'."

We'd like to take the opportunity to thank Dr Sally Corden, Dr Tom Connor and their teams for their hard work in securing more effective vaccines for Wales and their contribution to putting Welsh pathogen genomics on the world stage.

### 100,000 Genomes Project Achieves Sequencing Goal

The Welsh arm of the 100,000 Genomes Project has surpassed its recruitment goal for genetic sequencing.

The collective efforts of the All Wales Medical Genetic Service (AWMGS), Cardiff University, Wales Gene Park, Genomics England and the Welsh Government ensured that the project was brought to Wales after the success of its recruitment in England.

Since February, an incredible 439 participants and their relatives who are affected by undiagnosed rare genetic disorders have been recruited throughout Wales in an extremely short 8-month period.

DNA was extracted from the collected blood samples and subjected to Whole Genome Sequencing at the Sanger Institute. In addition, clinical information for the subsequent analysis was collected from all participants by November 2018.

By January 2019, initial results were returned for 68 of the 154 families. These results are currently being validated by the AWMGS laboratory.

Huge congratulations to all involved in the work; an important project that ensures that Welsh patients benefit from UK-wide initiatives.

## #JourneyTo Genomics

Showcasing just some of the many Genomics Partnership Wales members contributing to the future of Welsh Genomics

### What inspired you to pursue a role in the field of genomics?

I developed a new assay using the pyrosequencing platform for influenza resistance testing and have been interested in sequencing ever since. I have been able to assist my colleagues in many research projects throughout my career and when the opportunity came to develop new assays using forefront techniques as part of a new unit I relished at the challenge.

### What happens during a typical working day?

I work within a team with varying backgrounds. I am usually involved in the testing of Mycobacteria, HIV and Influenza. I prepare samples for Next Generation sequencing using Illumina reagents and run them on the MiSeq platform. I am enjoying every part of this new technique and I am amazed at the quantity and quality of the data we generate.

### What advice would you give to someone interested in joining the field?

To research the profession first and maybe ask to visit a laboratory to see what is involved. Some things can seem a bit mundane or repetitive and you need to make sure it is right for you before investing your time in the training and also to think about where you would like your career to go.



**Bree Gatica-Wilcox**

Specialist Biomedical Scientist  
Pathogen Genomics Unit (PenGU)

After graduating from University of Wales Institute, Cardiff with a degree in BSc Biomedical Science, Bree began working as a Biomedical Scientist in the Molecular Diagnostics Unit at Microbiology Cardiff (the former name of the Pathogen Genomics Unit) in 2003 before going on to obtain her MPhil in Medicine from Cardiff University in 2014.



**Dr. Marc Naven**

Bioinformatics Research Associate  
Wales Gene Park

Marc graduated from The University of Huddersfield with a degree in Medical Genetics in 2009 then completed his MSc in Bioinformatics at the University of Manchester in 2010. He gained his PhD from Cardiff University in 2015 for his body of work investigating therapeutic biomarkers in colorectal cancer before taking a post-doctoral role within Wales Gene Park.

### What inspired you to pursue a role in the field of genomics?

My family encouraged the scientist in me from a young age; I'd read puzzle books, build small electrical circuits and grow plants from seeds. Most of my spare time is spent doing those things today. Science fiction also had a big influence – I'm part of the original *Jurassic Park* generation.

### What happens during a typical working day?

I check the quality of all the data from the DNA sequencers – they generate loads, so this takes some time. The majority of my time is spent writing code and analysing the sequencing data. Then there are the project meetings and emails between the rest of my lab and collaborators.

### What advice would you give to someone interested in joining the field?

It's an exciting interdisciplinary field at the height of biotechnology; if you enjoy problem-solving, biology, computing, maths or chemistry you're bound to be rewarded by a job in genomics. There's also a bottleneck in the amount of skilled people in the field (especially bioinformaticians), meaning we're always in high demand!

**What inspired you to pursue a role in the field of genomics?**

Genomics in healthcare is a fast moving field, with constant developments. I like the challenge of using state of the art technologies and research to form practical services that directly impact patient’s lives.

**What happens during a typical working day?**

I am part of the Developmental Delay team. We perform genome wide analysis for children with intellectual disabilities and developmental delay. My average day includes investigation and reporting of, sometimes very unique, chromosomal abnormalities and interpreting how these might be contributing the patient’s condition.

**What advice would you give to someone interested in joining the field?**

Lab experience is really important to understand the underlying processes. Try to get some experience in laboratory research and organising projects where ever possible.



**Dr. Angharad Williams**  
Clinical Scientist  
*All Wales Medical Genetics Service*

A Cardiff University alumni, Angharad graduated with a BSc in Biochemistry and then a PhD. In 2014 she started working at AWMGS as a Pre-Registration Clinical Scientist, introducing new cancer biomarkers into clinical service. She recently completed training to become a registered Clinical Scientist in the Developmental Delay section.



**Eamonn Kirk**  
Registered Genetic Counsellor  
*All Wales Medical Genetics Service*

Eamonn graduated with a degree in Human Physiology, then went into the field of science publishing. He then switched to mental health work before gaining a Diploma and MSc in Psychiatric Social Work. After working with teenagers and young adults around issues including self harm, suicidality and sexuality, Eamonn moved into the field of genetics and has been a Genetic Counsellor for 11 years.

**What inspired you to pursue a role in the field of genomics?**

I have always liked the combination of working with people, where I have to use my head and my heart. I saw becoming a genetic counsellor as a chance to do this, and to make a difference to people’s lives. The field seemed dynamic and challenging, yet rewarding.

**What happens during a typical working day?**

I often have morning clinics. The rest of the day is made up of phone calls, letters, reviewing information, and meetings. The work is a nice balance between autonomy and team working. My favourite part is seeing clients and families.

**What advice would you give to someone interested in joining the field?**

I see the Genetic Counsellor role as vocational, so I think it’s important to want to make a difference. I would suggest someone hones their counselling skills, and develops good self-care strategies. And if interested, talk to someone who does the job!

*Tell the GPW community about your #JourneyToGenomics!  
Email Emma.Lynham@wales.nhs.uk for more information*



## Dates For Your Diary



### 'Pathogen Genomics: One Year On' Event

**14<sup>th</sup> February 2019, 10:00 – 15:30**

*The Life Sciences Hub, 3 Assembly Sq., CF10 4PL*

Celebrating the successful first year of the Pathogen Genomics Unit (PenGU), covering areas including computational biology, bioinformatives and next generation sequencing. Tickets are available from Eventbrite.



### Rare Disease Day Annual Reception

**14<sup>th</sup> February 2019, 18:00 – 20:00**

*The Senedd, CF10 4PZ*

Join Rare Disease UK for their annual Rare Disease Day reception at the Senedd, including an update on the Welsh Government Rare Disease Implementation Plan. Tickets are available from Eventbrite.



### GPW Genomics Ball

**19<sup>th</sup> July 2019**

*The Exchange Hotel, 4-5 Mount Stuart Sq. CF10 5FQ*

We're lauding the Welsh genomics revolution by throwing our first Genomics Partnership Wales Genomics Ball! Join us for an evening of celebration in the heart of Cardiff Bay. More details to follow.

## Contact GPW

@GenomicsWales

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## Programme Updates

### • GPW Brand Is Gathering Momentum

If you are attending any events or conferences, please let the GPW Programme Office know and we can support with merchandise. Follow us on Twitter and remind us of any stories that we can promote for you!

### • Medium Term Estates Solution Being Progressed

The need for new fit-for-purpose estates is ever-pressing. Partners are devising an integrated approach for the co-location of the AWMGS, PenGU and Wales Gene Park genomics teams. We're learning from other sites about how to work efficiently across disciplines to bring benefits to staff and patients. Want to know more? Feel free to contact the Programme Office.

### • Public and Patient Involvement Plans Underway

Do you know someone who has direct/indirect experience of genetic testing, or genetic conditions? We're recruiting for a Patient and Public Sounding Board to ensure that we learn from the experiences of those affected by genetics and genomics. Please visit <https://www.healthandcareresearch.gov.wales/current-opportunities/> for further information.

### • All-Wales Research Workshop Plans in Progress

Our aim is to bring researchers across Wales together for a one day workshop, to share their work and to discuss how we could enhance collaboration across Welsh Universities and academic institutions. Contact the Programme Office with any questions or to share your ideas.