

Foreword

Welcome to our first newsletter for 2020, later than planned but we are very happy to share our GPW news! The COVID-19 pandemic has needed the NHS and partner organisations to prioritise resources, thus in mid-March, we put the Strategy work on hold to allow this focus. The programme activity resumed at the end of June.

Despite the pause, a number of important services were introduced over the last few months; changes that will deliver real benefits to our patients - you can read about them in this issue.

These achievements are a great credit to the teams involved, and also reflect the benefit of partnership working and the investment provided by Welsh Government for the GPW programme. I would like to congratulate all of the partners involved.

We will build on these strong foundations to improve healthcare in Wales using genomics, keeping our patients as our main focus. We look forward to sharing this progress in future newsletters.



Michaela John
Programme Manager
Genomics Partnership
Wales

GPW in Focus

Wales leads the way in rapid sequencing diagnostics

The 'Welsh Infants' and Children's Genome Service (WINGS) for acutely unwell infants and children was launched by the All Wales Medical Genomics Service (AWMGS) in April. This involves looking at the whole genetic code (whole genome sequencing) of the child and both of their parents in order to detect whether there is a 'faulty' gene. Wales is the first UK nation to offer whole genome sequencing for seriously ill children.

Over the past year a multi-disciplinary team have developed this service in partnership - the AWMGS team alongside specialist Paediatric Consultants and Cardiff University colleagues. WINGS provides a faster diagnosis for some of the children, resulting in reduced hospital stays, more timely treatments and better outcomes. This service uses the NovaSeq, the high through-put sequencer introduced last year.

PenGU Update: Genome Sequencing For COVID-19 Pandemic Response

The Pathogen Genomics Unit (PenGU) has been working as a prominent part of the COVID-19 Genomics UK Consortium (COG-UK), PenGU have sequenced over 6,000 SARS-CoV-2 genomes. This places Wales third in the world for COVID-19 genomes sequenced.

The data collected is used to undertake outbreak investigations, support surveillance activities and provide evidence-based advice to Welsh and UK Government

Since early March, the PenGU team have built a cutting-edge COVID-19 genomics service that is delivering real benefit nationally. This achievement reflects the strength of longstanding collaborative links with Cardiff University and the investment that Welsh Government has provided as part of the Genomics for Precision Medicine Strategy.

What inspired you to pursue a role in genomics?

Back in 2004, while I was at the University of Liverpool, I worked with the then NHS chair for pharmacogenetics, on a project assessing the cost-effectiveness of HLA-B*5701 genotyping before prescribing Abacavir for patients with HIV. This test subsequently became standard of care, and has likely prevented many hundreds of cases of hypersensitivity syndrome.

What happens during a typical working day?

I have broad research interests, spanning pharmaceutical economics and policy through to medication adherence and pharmacogenetics. My research group is multidisciplinary with economists, mathematicians, and pharmacists and we use a number of methods from clinical trials, preference elicitation studies to pharmacometrics. The range of ideas and perspectives makes for highly rewarding research.

What advice would you give to those interested in joining this field?

Take a broad perspective, recognise the value of multidisciplinary, become a multidisciplinary. Practical advice would include getting training in complementary disciplines e.g. pharmacy and economics; pharmacology and genetics; medicine and statistics etc. Find a mentor.



Prof Dyfrig Hughes

Professor of
Pharmacoeconomics

Dyfrig qualified as a pharmacist before undertaking a PhD in pharmacology. He subsequently trained in health economics and is now Professor of Pharmacoeconomics at Bangor University. His research focuses primarily on the safe, effective and efficient use of medicines



Prof Steve Conlan

Professor of Molecular and Cell Biology; Head of Reproductive Biology & Gynaecological Oncology
Head of Enterprise and Innovation

Steve is a Programme Board member and Research Innovation lead for the GPW. He is also a founding director of Europe's first Centre for NanoHealth, a Fellow of the Royal Society of Biology, Trustee of the British Society of Nanomedicine, and honorary consultant in Swansea Bay NHS University Health Board

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

I 'discovered' genetics during A level Biology and have been hooked since! For my PhD, I sequenced half a gene, and during my first postdoc I used the first eukaryotic genome sequence i.e. *Saccharomyces cerevisiae*/brewers yeast extensively in my research. After completing a BSc in Biology I obtained my PhD from the University of London, and returned to a faculty position in Swansea University in 2000. Now I can't 'live' without genomics and the power it provides, especially for biomedical functional genomics, which is a big focus of what we do in my research group.

What happens during a typical working day?

I lead the Reproductive Biology and Gynaecological Oncology [RBGO] group at Swansea University; a vibrant multidisciplinary group using molecular and cell biology, as well as biophysics, nanotechnology and computer science (machine learning and AI) to tackle pressing medical problems. The favourite part of my day is to sit down with our young scientists and discuss their research findings.

What advice would you give to those interested in joining this field?

Genomics is a multidisciplinary subject, so you can do a degree in other disciplines. It is cutting edge science; and can be applied to research in medicine, evolution, environment and for animals, plants, and microbes. So if these are things that interest you, then Genomics could be for you too.

Programme Updates

Advancing Genomics Research in Wales with the Illumina NovaSeq 6000

Flexible and scalable, the NovaSeq 6000 will offer research facilities in Wales the capacity to conduct high throughput next generation sequencing studies in short time frames. The technology will additionally enhance the ability of research groups to continue to take part in internationally recognised, large-scale research projects such as comprehensive tumour profiling, biomarker identification and the development of novel therapeutic agents

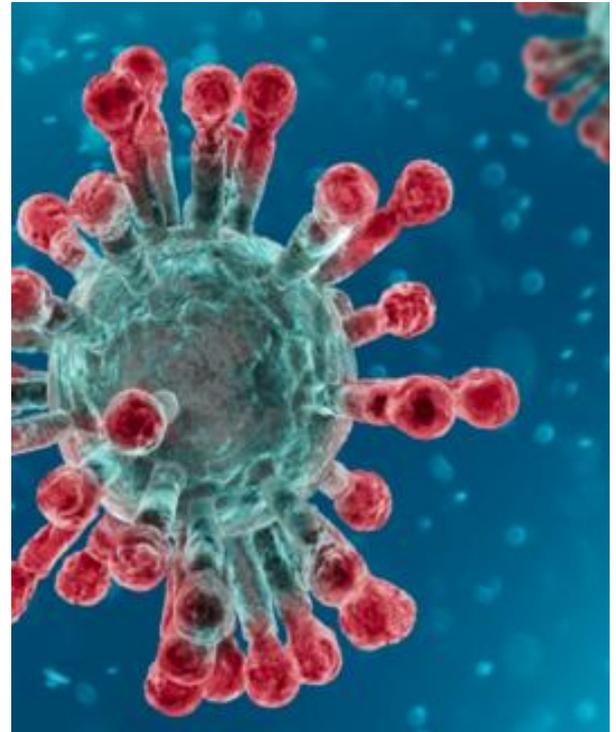
The field of genomics and precision medicine is rapidly evolving, our joint working approach as part of Genomics Partnership Wales (GPW) will ultimately enable research groups to undertake high quality research, which can translate to high quality clinical precision medicine services for both rare diseases and cancer genomics.

For more details, please contact the GPW Programme Office

Pharmacogenomics Service Introduced To Prevent Harm In Treating Cancer Patients

Following a local pilot, AWMGS have launched a national genetic screening service in Wales for patients undergoing fluoropyrimidine-based chemotherapy. The treatment is commonly used in the curative and palliative treatment for colorectal, oesophago-gastric and breast cancers and in some cases pancreatic cancer, hepato-biliary malignancies and malignancies of unknown primary site.

Although treatment with fluoropyrimidines is generally well tolerated, severe adverse drug reactions can result in death or escalation of care to high dependency and critical care units. A significant proportion of adverse drug reactions are as a result of individual genetic variation in the DPYD gene. Patients identified as having one or more copies of variants are considered for dose modification of fluoropyrimidines or use of alternative drug regimens.



COVID-19 (Coronavirus) UPDATE

It is encouraged that everyone should continue to do what they can to stop COVID-19 spreading. It is particularly important for the elderly, those who have a long-term condition, are pregnant or have a weakened immune system to take extra measures such as washing hands and social distancing as lockdown measures are eased

For the latest and most up to date information, visit the Public Health Wales website.

phw.nhs.wales

Programme Highlights

- **Genomics Cafés**

The Education & Engagement team at Wales Gene Park have been busy finding innovative ways to continue despite lockdown restrictions. The virtual cafés began in May and have been followed by COVID-themed cafés in July with further dates planned. Huge congratulations to the team!

- **Genomics Showcase**

Following the disappointing cancellation of our planned 2020 event, we are pleased to announce a two-day event in May 2021 – more information to follow in due course!

- **Estates**

The GPW plans to occupy the site at Coryton are underway. Requirements and initial designs are being developed at pace with agile and collaborative working being a key focus.

- **GPW Patient & Public Sounding Board**

On 14th July we inducted eight new members to our Sounding Board, bringing the total number of members to 17. We are currently holding our Sounding Board consultations virtually to ensure that we continue to co-produce our strategy despite the pandemic restrictions.

- **Covid-19 Research and Innovation**

In addition to the introduction of key services detailed in this edition, GPW has made significant contributions to research and innovation throughout the pandemic. A wide range of partners have been recognised for their contribution in a paper published in The Lancet which can be read here:

genomicspartnership.wales/resources

- **Do you have a good news story to share? Send us an email to be featured in the next issue.**

And Finally...

We are pleased to announce that our website is now live! The site will be home to our latest updates, events and resources from GPW and its partners. Visit GenomicsPartnership.Wales to find out more.



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Working together to harness the potential of genomics to improve the health, wellbeing and prosperity of Wales.

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