Wales Gene Park support for the Welsh Governments “Genomics for precision medicine” strategy
“To promote and facilitate Welsh medical genetic and genomic research and its application to improve health and wealth in Wales. Also, to engage the public and health professionals to improve understanding of the opportunities and challenges arising through genetics and genomics.”

**Mission Statement**

- **Genomics**
  - The study of an organism’s complete set of genetic information.
  - ‘Genome’ - the complete genetic information of an organism.
  - The genome includes both genes and non-coding DNA.

- **Genetics**
  - The study of heredity
  - The study of the function and composition of single genes.
  - ‘Gene’ - specific sequence of DNA which codes for a functional molecule.

Taken from @genomicsedu
Work package 1
Genomic Editing & Transgenics
- Genome Editing Lead Prof. Ros John
- Transgenic Pre-clinical Trials Lead Dr Ming Shen
- Genome Editing Research Manager Mrs Bridget Allen
- Research Associate Dr Jian Yang
- Research Associate Dr Kalin Narov

Work package 2
Genomic Facility
- NGS Laboratory Lead Ms Sarah Edkins
- Bioinformatics Lead Dr Kevin Ashelford
- Research Manager Ms Shelley Rundle
- Bioinformatician Dr Marc Naven
- Research Technician Dr Vikki Humphreys
- Bioinformatician Dr Anna Evans
- Research Technician Mrs Jincy Winston
- Bioinformatician Dr Peter Giles WCRC funded

Work package 3
Education and Engagement
- Work-Package Lead Mrs Angela Burgess
- Education and Engagement Officer Dr Rhian Morgan
- Genetic Alliance Policy & Engagement manager Wales Mrs Emma Hughes
- Education assistant Mrs Nina Lazarou

Technical and research coordination support
- Research Technician Julie Maynard
- Research Coordinator Dr Hala Jundi
- Research Coordinator TBA
- Research Associate Dr Anna Derrick

UKCRN project governance
Neurology & Molecular Neuroscience group - Swansea Neurology Biobank
Researchers provide:
The idea!
The samples
Consumable costs

Collaboration

Wales Gene Park provide:
Expertise
Sequencing
Bioinformatics
Training

Research

All supported by Welsh Government through Health and Care
WGP impact

- Supporting Research across Wales
  - Since April 2015 (current funding period)
    - 73 research grants supported, and £20,706,837 grants awarded
    - Supported 170 research articles

- Upskilling health professionals and informing the public
  - A total of 7178 people attended WGP Education & Engagement events in 2016
  - 1450 Health professionals across Wales received training

- Informing and implementing Government Policy
  - Welsh Rare Disease Implementation Plan (revised July 2017)
  - Welsh Government Genomics for Precision Medicine Strategy (launched July 2017)
Genomics for Precision Medicine Strategy

Last updated 06 July 2017

Our plan to create a sustainable, competitive environment for genetics and genomics to improve health and healthcare provision for the people of Wales.

Precision Medicine is an emerging approach for disease treatment and prevention that takes into account an individual's variability in genes, environment, lifestyles etc.

Rare diseases implementation plan

Last updated 31 July 2017

A rare disease is defined as a life-threatening or chronically debilitating disease that affects five people or less per 10,000.

There are around 150,000 people affected by these diseases in Wales, most are genetic.
Rare Disease UK & the Welsh Rare Disease Patient network: Empowering patients to take part in the policy process

In collaboration with the Wales Gene Park, RDUK established the Welsh Rare Disease Patient Network to engage patients, families and patient organisations to ensure the patient voice is properly informed of, and effectively represented in the discussion and development of the implementation of the UK Strategy for Rare Diseases. The network was launched in October 2015. Over 80 people attended the launch and signed up to the network. Contact is maintained with network members through email and a twice yearly newsletter keeping them informed of latest news, advances and initiatives. Patient involvement in events such as Rare Disease Day, Patient Pledge Campaign & a Twitter Takeover has been enhanced by using network contacts. The network continues to grow with over 140 members at present.

'Working collaboratively with other groups as part of the rare disease patient network provides a united voice for campaigning to improve rare disease provision for patients and families across Wales'.
Kayleigh Old, Public Affairs Officer, Cystic Fibrosis Trust
Ensuring patient voice is heard and acted upon
  o Co-ordinate the Welsh Rare Disease Patient Network

Facilitating patient recruitment into Research
  o A Welsh Genomic Medicine Centre is to be developed initiated through public participation in 100K genomes project in Wales
Committed to support AWMGS to develop model of **informed patient consent** to allow routine research access to surplus clinical genetics samples and data for medicine and genomics research.

Training Health and Social Care Professional
- Ensuring better identification of rare diseases to deliver faster diagnosis and access to treatments &/or support for patients.
- Mainstreaming genomics into the wider NHS

Continue to promote genetic literacy and raise awareness of genomics within the public in Wales

All Wales Medical Genetics Service
WGP supporting policy implementation

- Development of IT infrastructure, networking capabilities and data governance for NHS in Wales.
  - Establishing a two-way secure network between NHS and CU
  - Working with the SAIL databank to link genomic data with other routinely collected health/social data
  - ISO accreditation within CU data centre

- Knowledge transfer into the NHS for the benefit of service delivery
  - NHS representation within the College research data strategy
  - CU representation within the NHS IT implementation group
Lesley Roberts was extremely positive about the holistic package of care that was offered by the multi-disciplinary team at Cardiff - offering clinical expertise, research opportunities and provision of information and support through coordination with an advisor from the Tuberous Sclerosis Association based at the clinic.

TSC gives rise to non-cancerous growths in various organs, that can lead to severe complications. Patients affected by TSC often require expertise from a number of specialties.

Provide a multi-disciplinary and coordinated approach to management and treatment for patients with this rare condition.

Offers opportunities for patients to contribute to research.

Translation of pre-clinical research into using mTOR inhibitor treatment (Everolimus), is now being prescribed for patients with TSC tumours in the specialist TSC clinic.
Contacts

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