Partneriaeth Genomeg Cymru
Genomics Partnership Wales

Poster Abstracts

1

Title - Optimising Glaucoma Pathways in NHS Wales: Integrating Genomic Risk Stratification through a Quality Improvement Approach

Authors - ***Deepak Shaji Senior Project Support Officer Ophthalmology Directorate, Cardiff & Vale University Health Board***

Abstract - Glaucoma remains a leading cause of irreversible blindness in Wales, with outcomes closely linked to timely detection and consistent monitoring. Emerging genomic evidence highlights inherited susceptibility and risk-associated variants that may enable earlier identification of individuals at increased risk. However, the practical integration of genomic insight into routine ophthalmology services remains limited. This service-focused perspective explores how genomic risk stratification could be embedded within glaucoma pathways in NHS Wales using structured quality improvement methodology. By analysing existing referral, triage, and follow-up processes, opportunities can be identified to align genetic risk indicators with clinical decision-making, surveillance intervals, and patient communication. Integrating genomic data alongside established parameters—such as intraocular pressure, optic nerve assessment, and visual field analysis—may support more personalised and preventative models of care. Applying improvement principles ensures that innovation is implemented safely, equitably, and sustainably. The strategic incorporation of genomics within glaucoma services has the potential to enhance early intervention, reduce avoidable progression, and optimise resource allocation as precision medicine becomes increasingly embedded across NHS Wales.

2

Title - Cracking the (Genomic) Code: A Mixed Methods Exploration of Genomic Knowledge, Practice, and Implementation in Nursing and Midwifery

Authors - **†Thomas, J., *Tonkin, E., *Lancastle, D., *Davies, M., †‡Murray, A., ‡John, M. *University of South Wales, United Kingdom †All Wales Medical Genomics Service, United Kingdom ‡Genomics Partnership Wales, United Kingdom*

Abstract - Advances in genomics offer opportunities for enhanced diagnostics, risk stratified screening, and targeted therapies for the people of Wales. Achieving this requires a workforce with the knowledge and confidence to embed genomics into routine care. As the largest professional group in the NHS, nurses and midwives are central to delivering genomic healthcare. This doctoral research project explored how genomics is integrated across nursing and midwifery education and practice using a convergent mixed methods design: a scoping literature review, a survey of genomic knowledge among pre and postregistration healthcare students, and qualitative interviews with nurses and midwives using genomics in their roles. Findings revealed significant gaps across the education–practice continuum. The literature showed growing global engagement of nurses and midwives in genomics, but implementation was fragmented. Students demonstrated limited genomic knowledge and minimal exposure in educational and clinical settings. Practising nurses and midwives viewed genomics as separate from traditional professional knowledge, with engagement constrained by systemic barriers. Synthesising these findings led to an original conceptual model describing a cyclical set of factors inhibiting genomic integration in nursing and midwifery. Strength based recommendations were developed to address these factors and support efforts to enhance genomic capability among the nursing and midwifery workforce in Wales.

3

Title - The TSC2 Transcriptional Activation Domain is Associated with Partially mTORC1-Independent Transcriptional Changes Involving Extracellular and Inflammatory Gene Programmes.

Authors - *Samia S. Alzahrani*, Jesse D. Champion*, Darius McPhail*, Mohammad A. M. Alzahrani*, Brian L. Calver*, Elaine Dunlop*, Mark Davies*, Andrew R. Tee*. *Division of Cancer and Genetics, Cardiff University, Heath Park, Cardiff CF14 4XN, UK.*

Abstract - Tuberous sclerosis complex (TSC) is a genetic disorder caused by loss-of-function mutations in TSC1/TSC2 leading to multisystem tumour formation. Although inhibition of mTORC1 reduces tumour growth, clinical manifestations frequently persist, suggesting the involvement of additional mTORC1-independent mechanisms. Emerging evidence suggests that TSC2 contributes to transcriptional regulation beyond canonical growth signalling. Transcriptomic and functional analysis were performed in engineered TSC2 models. Transcriptomic analysis identified coordinated alterations in extracellular matrix, inflammatory, and signalling-associated gene networks, including FN1 and multiple collagen-related genes. mTORC1 inhibition resulted in only partial normalisation of these transcriptional changes, indicating persistence of a subset of gene expression alterations independent of mTORC1 activity. Comparative analysis between GAP- and TA-domain mutants revealed distinct transcriptional profiles, with TA-domain disruption associated with altered expression of genes linked to extracellular signalling and microenvironmental interactions. STAT3 inhibition partially modulated a subset of this transcriptional signature, whereas rapamycin showed comparatively limited transcriptional effects. These findings suggest that TSC2 loss is associated with mTORC1-idependent transcriptional alterations involving extracellular and inflammatory gene programmes, supporting regulatory roles beyond mTORC1 signalling.

Title - Driving National Collaboration in Advanced Therapy Research and Delivery

4

Authors - *Advanced Therapies Wales - Lin Amor*

Abstract - Advanced Therapies Wales is the only programme of its kind in the UK, providing a national focal point for expertise & support. Established in 2019 with a statement of intent from Welsh Government it has become a central point for resources, clinical activity & industry ambition. It offers a one-Wales approach with a unified strategy to enable patient access. The Delivery Plan for Advanced Therapies in Wales was written in 2024 on behalf of the Welsh advanced therapies ecosystem, with its delivery coordinated & facilitated through the ATW Programme. It captured the steps required to progress the development of the advanced therapies sector across Wales, over the subsequent five years ensuring alignment with National policies, objectives and ambitions. The Delivery Plan is centred around 4 key goals and 3 cross cutting themes. There has been significant progress for the advanced therapies sector in Wales over the last year, driven by a shared commitment to innovation, equity, and collaboration. Wales has moved from participating to leading.

5

Title - How patient and public voices are integral to our work

Authors - *Midlands-Wales ATTC/ATW - Lin Amor NHS Wales JCC- Hannah Crocker Midlands-Wales ATTC/ATW - Rebecca Curtis Midlands-Wales ATTC/ATW - John Lewis*

Abstract - Midlands-Wales Advanced Therapies Treatment Centre (MW-ATTC) are part of the Advanced Therapies Treatment Centre Network (ATTC) who address the unique and complex challenges of bringing advanced therapy medicinal products (ATMPs) to patients. Advanced Therapies Wales (ATW) is the only programme of its kind in the UK, providing a national focal point for ATMP expertise and support, and co-lead the MW-ATTC. Friends of Cymru Sickle Cell and Thalassaemia (FOCST) invited us to attend their community education and engagement event. The aim was to build a framework of knowledge about two new advanced therapies which were soon to be available in standard care. By bringing together patients, clinical experts, clinical psychologists, carers and members of supporting voluntary organisations the hope was to find a shared language, discuss issues of importance and learn more. People with lived experience and their loved ones asked questions and shared their stories with us. We took away key messages which we integrated into our work: We formulated the agenda for our annual symposium event to ensure patient voices were at the fore; We co-produced a new ATW website with patient and public representatives ensuring information was presented in clear language in an accessible format. We also formulated a list of current and future projects to develop this workstream further.

6

Title - Optimising Pre-Cardioversion Management: A Quality Improvement Audit of Anti-Arrhythmic Therapy Use in Outpatient DC Cardioversion

Authors - *Dr Kerolos Nakhnokh (Senior clinical fellow) Betsi Cadwaladr University Health Board, Wales, UK Dr. Royle Martin (Cardiology consultant) Betsi Cadwaladr University Health Board, Wales, UK Jasmine Doble (Physician associate) Betsi Cadwaladr University Health Board, Wales, UK*

Abstract - Direct current (DC) cardioversion is an established rhythm control strategy for atrial fibrillation and atrial flutter, yet patient selection and optimisation remain variable. This retrospective quality improvement project evaluated outpatient DC cardioversion outcomes from May 2024 to January 2025, focusing on the impact of pre procedure antiarrhythmic therapy. Eighty-six patients (mean age 70 years) were analysed; 41 received antiarrhythmic drugs other than betablockers. Success rates were similar between treated and untreated groups (91.7% vs 88.9%), with no statistically significant difference (χ^2 p=0.33). At four week follow up, sinus rhythm maintenance was 59.3% overall, again with no significant difference between groups (p=0.66). High early recurrence and inconsistent pre procedure optimisation highlighted key gaps, including absence of a standardised assessment pathway. The project led to development of a structured pre cardioversion checklist covering rhythm history, comorbidities, thyroid status, medication optimisation and echocardiographic factors. Future implementation aims to improve patient selection, reduce recurrence, and support more consistent clinical decision making across services.

7

Title - The Wales Cancer Biobank: Evolution of bio resourcing to serve both patients and the evolving multimodal appetite of the cancer research community in Wales

Authors - *(1) Liz Merrifield, (2) Dr. Lisa Spary, (2) Abigail Macarthur, (3) Mr Daniel Naeh, (1) Dr. Peter Giles, (2) Prof. Richard Adams, (2) Prof. Richard Clarkson 1. BioResource Data Accelerator, Wales Gene Park, CardiN University 2. Wales Cancer Biobank, CardiN University 3. Wales Cancer Biobank, Swansea University*

Abstract - The WCB holds extensive cancer sample archives collected over 20 years. Its approval and consent model enables direct linkage between biosamples and patients' NHS e health records, creating richly linked clinical–biological datasets for research. With rapid advances in AI, big data, and genomics transforming cancer research, biobanks must evolve to meet new data needs. Therefore, WCB are therefore responding to this by developing Wales' first national Bioresource, expanding the multimodal data available to researchers. Biological sample provision remains WCB's core function, distinguishing it from other national biobanks that limit sample access. Several projects are enhancing WCB's cancer data infrastructure, including establishing pipelines to genomic diagnostic data via AWMGS, creating a histopathology image databank to support AI diagnostics, and acquiring radiological imaging datasets. Strong PPIE initiatives are modernising (digitising) consenting processes through iConsenting, with plans to broaden these digital consent procedures nationally. Together, these developments demonstrate WCB's collaborative approach with academic, NHS, and industry partners, supporting research aligned with CReSt priority themes and delivering improved resources for cancer patients.

8

Title - The BioResource Data Accelerator: An exciting update from this all-Wales approach to sample and genomic data access for the cancer research community.

Authors - *(1) Liz Merrifield, (1) Dr. Peter Giles, (2) Dr Andrew Fry, (2) Rhys Vaughan, (3) Prof. Richard Clarkson, (3, 5) Prof Richard Adams, (5) Angela Casbard, (4), Sian Morgan, (4) Dr. Joseph Halstead, (4) Dr Sophie Shaw, (6) Jennifer Selby, Dr. Joanna Zabkiewicz (2) 1 = BioResource Data Accelerator, Cardiff University 2 = Wales Gene Park, Cardiff University 3 = Wales Cancer Biobank, Cardiff University 4 = All Wales Medical Genomics Service 5 = Centre for Trials Research, Cardiff University 6 = National Data Resource, Digital Health and Care Wales*

Abstract - Since 2022 the BioResource Data Accelerator (BRDA) has supported major initiatives in cancer & rare inherited diseases. In 2025, the programme advanced data utility for rare disease & cancer research & healthcare, aligning with key national strategies & incentives. Welsh National Bioresources: BRDA continued developing the Wales Cancer Biobank (WCB) into a national bioresource, including new reproducible analytical pipelines with the All Wales Medical Genomics Service (AWGMS). These improvements expand access to genomic healthcare records linked to donated biosamples & diagnostic data, & now look to establish similar pipelines of data access for radiological image data for the WCB together with the National Imaging Academy Wales. We also supported the creation of the All Wales Genomic Databank for rare inherited disease patients. Enhancing Secure Data Environments: Functionality in the National Data Resource (NDR) and SAIL Databank increased through two projects. The NDR enabled control cohort analysis for the QuicDNA lung cancer trial, comparing data quality & availability between standard-of-care & trial participants; and through the successful upload of AWGMS VCF data into SAIL, enabling population-level genomic linkage. Lynch syndrome data integration is planned for 2026. Strategic leadership: Our team are leading the strategic vision for the Cardiff Cancer Research Partnership, supporting early phase trials & efforts to improve patient experience across cancer pathways.

9

Title - Cri-Du-Chat syndrome in neonate - A case report

Authors - *PAL R 1, Dave T 2 1 Hywel Dda Health Board, Carmarthen Carmarthenshire, United Kingdom 2 Hywel Dda Health Board, Carmarthen Carmarthenshire, United Kingdom*

Abstract - Background: Cri du Chat syndrome is caused by a partial deletion on the tip of the short-arm of chromosome 5. Characterised by cat like cry in neonatal period and multiple comorbidities including Craniofacial malformations, developmental and behavioural manifestations, profound learning difficulties.

Aim: We present a case of female infant with de novo mutation who is now over two years old. The aim of the report is to explain the journey and challenges faced by the infant and family. Born at 37 weeks of gestation and had respiratory distress initially for which was admitted to NICU. Chest X-ray reported 11 pair of ribs along with distinctive facial features. SNP array identified two chromosome changes- 5p microdeletion and Xq28 microduplication. The Xq28 duplication had attached itself to the end of chromosome 5 (the copy of which has the deletion), resulting in unbalanced chromosome translocation. In our patient's case these changes have resulted in - Delayed milestone, feeding difficulties, trigger finger of left hand, small stature, recurrent LRTI due to frank aspiration, macroglossia, obstructive sleep apnoea, upper lip tie and high arch palate.

Conclusion: The patient has been catching up on developmental milestones with multidisciplinary support, gaining weight steadily on appropriate diet after gastrostomy and on prophylactic antibiotics to keep infections under control. Her prognosis remains well at this point with regular care and multidisciplinary follow up.

10

Title - Lynch syndrome in Wales: insights from routine health data adapted for surveillance and screening

Authors - *Robert Maddison (1, 2), Ceri Williams (1), David Tucker (1), Suki Baynton (1), Sian Nisbet (3), Mark Davies (4), Alex Murray (3), Llion Davies (1) 1. Congenital Anomaly Register and Information Service, Public Health Wales, Capital Quarter 2, Tyndall Street, Cardiff, CF10 4BZ 2. Wales Gene Park, Cardiff University, Wales Genomic Health Centre, Cardiff Edge Life Sciences Park, Longwood Drive, Whitchurch, Cardiff, CF14 7YU 3. All-Wales Medical Genomics Service, Wales Genomic Health Centre, Cardiff Edge Life Sciences Park, Longwood Drive, Whitchurch, Cardiff, CF14 7YU 4. Singleton Hospital, Sketty Lane, Sketty, Swansea, SA2 8QA, UK*

Abstract – Background: Lynch syndrome (LS) is an inherited condition caused by pathogenic variants in the mismatch repair genes: MLH1, MSH2, MSH6 and PMS2. LS greatly increases the risk of developing a variety of cancers, making regular screening key for early diagnosis and treatment. Effective patient registration to support such screening is currently lacking in Wales.
Aims: We will assess the feasibility of developing a Welsh LS register using routine health data for surveillance and screening purposes.

Methods: Data collection is ongoing. LS patients will be identified through two routes: 1) querying young (<50 years) specific cancer cases in the Patient Episode Database for Wales, and 2) using LS genetic test reports held by the All-Wales Medical Genomics Service. These reports will be used to positively identify LS patients who may be considered for screening. Descriptive epidemiology analyses will be undertaken.

Results: We will evaluate the strengths and limitations of the data sources used to construct the register and report insights for the future monitoring of cancer predisposition syndromes. We anticipate that linking genetic test reports will substantially enhance the identification of LS patients who are likely to benefit from screening.
Conclusion: By exploring registration of LS patients from routine data, we aim to improve screening efficacy and early detection for LS-associated cancers in Wales.

11

Title - Developing the Genomic Medicine Workforce: Supporting Healthcare Professionals in Part-Time Postgraduate Study

Authors - *Jodie Ann Croxall and Claire Morgan. Faculty of Medicine, Health and Life Sciences. Swansea University. Swansea. SA2 8PP*

Abstract - Background: Rapidly advancing disciplines such as genomic medicine depend on healthcare professionals accessing postgraduate education while remaining in clinical practice. However, postgraduate systems are traditionally structured around full-time learners, potentially creating misalignment for working professionals.

Aim: To explore how part-time postgraduate study can be supported for healthcare professionals in Wales and identify structural factors shaping participation.

Methods: A cross-Wales mixed-methods online survey of healthcare professionals enrolled in part-time postgraduate programmes was conducted. Quantitative responses were analysed descriptively and qualitative responses were reviewed thematically.

Results: Participants reported pressures associated with combining study and employment, including limited access to protected study leave, reliance on annual leave, workload pressures and restricted access to university resources outside standard hours. Conversely, employer support, constructive educator relationships, peer learning communities and flexible or blended learning models supported participation.

Conclusions: Developing a genomics-ready workforce requires stronger alignment between universities and healthcare employers. Protected study leave, improved access to digital resources, employer–university partnerships and flexible programme design may support healthcare professionals undertaking postgraduate education alongside clinical roles.

12

Title - Investigating the distribution and frequency of CFTR alleles in Wales using linked routine genetic data

Authors - *Robert Maddison 1, Karen Reed 1, Rebecca Cannings-John 2, Fiona Lugg-Widger 2, Thomas Stoneman 3, Sarah Anderson 3 ,Andrew E. Fry 1,3 1. Wales Gene Park, Division of Cancer and Genetics, Cardiff University, Cardiff, CF14 4XN, UK 2. Centre for Trials Research, Cardiff University, Cardiff, CF14 4XN, UK 3. All Wales Medical Genomics Service, University Hospital of Wales, Heath Park, Cardiff, CF14 4XW, UK*

Abstract - Background: Routinely collected clinical genetic test records are underutilised for research. We aimed to explore the utility of such records for population genetics by linking cystic fibrosis (CF) genetic test reports to demographic datasets in the SAIL Databank, a trusted research environment (TRE).

Methods: We curated and imported 16,181 CFTR gene test reports from AWMGS to the SAIL Databank for anonymisation. Spatial mapping was performed with respect to allelic diversity, rurality and Welsh language. CFTR allele frequencies were compared with estimates from 1993. Linked demographic data were used to detect related individuals who might impact the frequency estimate.

Results: CFTR alleles appear distributed in a rural-urban manner, with highest carriage rates and diversity in South Wales, though this may be due to differences in regional service utilisation. RCGD captures mild and severe alleles with greater sensitivity than prior registry based methods. Linked census and birth records were used to infer relationships to increase allele frequency estimate accuracy. Frequencies could not be reported safely due to disclosure risk from small numbers.

Conclusion: Linkage of routine clinical genetic test records can facilitate population genetics research, providing new insights, but our experience has highlighted limitations which have particular implications for rare disease research: reporting limitations, undetected related individuals and ascertainment bias.

13

Title - HSP90AA1 as a Novel Candidate Therapeutic Target in the regulation of EGFR following the loss of Kidins220 using RNA sequencing analysis in pancreatic cancer cells

Authors - Deepa Shankla1, Qingping Dou2, Wen G. Jiang1 and Lin Ye1 1 Cardiff China Medical Research Collaborative, Division of Cancer and Genetics, Cardiff University School of Medicine, Cardiff, CF14 4XN, UK. 2 Barbara Ann Karmanos Cancer Institute, Departments of Oncology, Pharmacology and Pathology, School of Medicine, Wayne State University, Detroit, MI 48201, USA

Abstract - Introduction: The molecular chaperone HSP90AA1 is essential for protein folding and maintenance of cellular homeostasis. Our previous studies in pancreatic cancer demonstrated that knockdown of Kidins220 leads to increased EGFR expression; however, the underlying regulatory mechanisms remain unclear. This study aimed to investigate the role of HSP90AA1 in EGFR regulation following Kidins220 knockdown.

Methods: Kidins220 was silenced using lentiviral shRNA in pancreatic cancer cell lines MiaPaCa-2, PANC-1, and AsPC-1, with scramble controls. RNA sequencing was performed to identify transcriptional changes associated with Kidins220 loss. Knockdown efficiency was confirmed by PCR, qPCR, and western blot. Cells were treated with an HSP90 inhibitor (50 nM) for 2, 4, and 24 hours, and EGFR protein levels were analysed by western blot. Invasive capacity was evaluated using a Matrigel invasion assay.

Results: Kidins220 knockdown resulted in increased HSP90AA1 expression across all three cell lines. HSP90 inhibition altered EGFR expression, with elevated levels observed at 24 hours compared with untreated controls. Additionally, invasion assays demonstrated reduced invasive potential in knockdown cells.

Conclusion: These findings suggest that HSP90AA1 may contribute to EGFR regulation following Kidins220 knockdown and could represent a potential therapeutic target in pancreatic cancer

14

Title - The role of the gut microbiota in patient response to chemotherapy.

Authors - *Morgan, C, Lalwani, N., Adegun, T., Harris, D and Facey, P.D. Swansea University Medical School, Faculty of Medicine, Health and Life Sciences, Singleton Park Campus, Swansea. SA2 8PP*

Abstract - Growing evidence suggests that the human gut microbiota influences cancer risk by promoting tumorigenesis. Additionally, these microbes may also alter patient response to chemotherapy. Evidence is now mounting that some bacteria in the gut microbiota may have the ability to alter chemotherapeutic drugs, thus reducing treatment efficacy. However, which species can do this is largely underexplored. Our research investigates not only how a patient's own gut microbiota may affect treatment response but also which microbial genes are involved. Rather than screening for the next oncopathogen, this study aims to map chemotherapy-modifying microbial genes across the bacterial kingdom. While others focus on microbes considered pathogenic, our preliminary genomic analyses indicate that these genes also occur in species considered beneficial. This research highlights that previous unconsidered bacteria may also affect patient responses to chemotherapy. Furthermore, we argue that microbial screening alongside existing tests, such as the faecal immunochemical test (FIT), should be implemented. By identifying chemotherapy-modifying bacterial genes, this research aims to improve the understanding of how the gut microbiota shapes chemotherapy response, thus improving outcomes for patients with colorectal cancer.

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Title - Evaluation of the Feasibility of Implementing Single Nucleotide Polymorphism Array Testing in Patients with Myelodysplastic Syndrome and Development of the Testing Pathway.

Authors - *Williams, M All Wales Medical Genomics Service, Cardiff, UK*

Abstract - Cytogenetics is essential for diagnosis, prognosis and treatment selection in patients with myelodysplastic syndrome. At AWMGS, conventional karyotyping has become unsustainable due to increased demand and workforce shortages, resulting in prolonged turnaround times. Single nucleotide polymorphism arrays offer high-resolution, culture-independent genomic profiling and may offer a more efficient alternative. Aim to evaluate performance of MDS karyotyping and SNP array services, compare cytogenetic findings and assess operational and clinical feasibility. A series of service evaluations were performed, including retrospective review of two years of MDS karyotyping referrals, technical review of SNP array service, concordance analysis of 30 samples, and operational modelling. Findings informed a quality initiative including risk assessment, stakeholder engagement, development of documentation, training and reporting workflows, and implementation plan. Karyotyping had a 114-day average turnaround time with 9% of samples meeting the national 21-day target. SNP arrays show high analytical quality, reduced staff analysis time and improved detection of sub microscopic abnormalities. Concordance analysis demonstrated detection of major unbalanced abnormalities relevant to risk stratification and expected limitations mitigated through revision of sample pathways. Overall, SNP arrays were deemed to be clinically beneficial and operationally feasible as first-line cytogenetic test for MDS.

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Title - Beyond the Human Genome: Understanding the Microbiome's Role in Cancer Care

Authors - *Dr Paul Facey, Natasha Lalwani and Dr Claire Morgan. Faculty of Medicine, Health and Life Sciences, Swansea University. SA2 8PP*

Abstract - Genomics usually focuses on human DNA, but we also coexist with trillions of microorganisms whose collective genetic material—the microbiome—profoundly influences health. Advances in DNA sequencing now allow scientists to study not only tumour genetics but also the bacterial genes living within us. Emerging research suggests the microbiome may influence cancer development, responses to chemotherapy and immunotherapy, and treatment-related side effects. Certain bacterial communities have been associated with improved treatment outcomes, while others are thought to affect inflammation and drug metabolism. While still an evolving field, this raises a vital question: could microbiome profiling help personalise cancer care in Wales? This educational poster aims to support public understanding of microbiome science in the context of cancer care. It introduces how microbial DNA can be collected, sequenced and analysed alongside tumour genomic data. The poster also considers why awareness and engagement are important as this field develops. Understanding how new genomic approaches may influence personalised treatment pathways raises questions around sample donation, data use and expectations of benefit. As research continues to explore interactions between human and microbial genetics, improving public understanding will be essential to ensure that future microbiome-informed cancer care is developed in ways that are ethical, inclusive and responsive to patient needs.

17

Title - Facilitating Timely Access to Genomic Medicines Through the New Treatment Fund: Insights From 2025

Authors - *Levey, C., *Francis, S., *Haines, D., *Adams, H. *All Wales Therapeutics and Toxicology Services

Abstract - Ensuring access to medicines that require genomic testing for patient eligibility depends on the NHS in Wales being prepared to deliver these services. The New Treatment Fund (NTF) aims to improve timely patient access to medicines recommended by AWMSG or NICE. The NTF start date should align with appraisal approval, and the NHS in Wales should make the medicines available within 60 days. However, national delays to the NTF start date can occur due to the unavailability of new testing services. All medicines recommended by NICE and AWMSG between January and December 2025 were reviewed. A medicine was classed as requiring a genomic test if NICE or the manufacturer indicated a testing requirement. Implementation time was measured from appraisal approval to NHS providers in Wales confirming NTF availability. Of 71 medicines recommended in 2025, 14 (20%) required a genomic test. Of these, twelve medicines (86%) had no national testing delays, and had a median implementation time of 13 days (0 – 113 days). Two medicines had national delays due to the issues in delivering the required genomic/pathology service. Median implementation times by NHS providers for these medicines were 39 and 50 days respectively. Most medicines requiring genomic testing had no national testing delays, and the medicines with testing delays were still implemented within 60 days. This suggests that genomic/pathology services could be effectively delivered.

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Title - Enhancing Genomics in Medical Education Through E-Learning Resources

Authors - Hannah D. West1, Devon Ward2, Zuzanna Grygiel1, Karen Reed1, Keith Hart1, Elaine A. Dunlop1 1. School of Medicine, Cardiff University, Cardiff, CF14 4XN, UK 2. Liverpool University Hospitals Group, Liverpool, UK

Abstract - With increasing integration of genomics into healthcare, embedding genomics within undergraduate medical curricula helps prepare future clinicians for personalised approaches to diagnosis and treatment. As many students find genomics challenging, we developed e-learning resources aligned to Years 1–3 of the Cardiff University MBBCh curriculum to improve confidence and engagement. These resources support case-based learning, contextualise key concepts and show the relevance of genomic medicine to current clinical practice. Our Genomics Lightbulb resources link to Year 2 case scenarios, helping students recap core genetic concepts and understand how genomics applies across specialties. To illustrate how genomics informs oncology, we created a virtual breast cancer patient pathway where students make decisions at key points to determine the final journey. To incorporate lived experience, we produced podcasts featuring individuals affected by genomic conditions, who discuss decision making, navigating inherited disease and advocating for themselves and their families. Used in Year 3 Rare Disease Week, these narratives provide insights into the ethical, emotional and social dimensions of genomics that are often absent from traditional teaching. By integrating this flexible digital learning with real world perspectives, these resources help develop genomics-aware clinicians ready to use genomic information in practice.

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Title - CCRNavigator: An Interactive Tool for Visualizing Constraint Coding Regions and Single-Dataset VCFs in hg38

Authors - Tyler Adams, Cardiff University, United Kingdom

Abstract - Constraint Coding Regions (CCRs) highlight segments of the genome with reduced tolerance to functional variation and are useful for interpreting rare variants. To support focused exploration of these regions, we developed CCRNavigator, an R/Shiny application that provides an interactive view of CCR90/95/99 intervals together with a single user selected, indexed VCF dataset. The application uses GRanges based subsetting and Gviz multitrack visualization to display CCRs and variants within a user defined genomic window. Users can pan, zoom, and recenter the view, allowing inspection at scales ranging from tens to thousands of base pairs. Variant positions and IDs can be retrieved through a simple click based interface, and the single dataset design keeps the workflow straightforward when working with large or individual VCF files. CCRNavigator offers a compact, reproducible environment for examining CCRs and visualizing variant context within the hg38 reference genome, supporting research tasks that require fine scale inspection of constrained regions.

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Title - The Literature Review Predating A Grant Proposal for the Development of a Gene Therapy for Wilms' Tumour Suppressor Gene Mutations causing Denys-Drash Syndrome

Authors - *1Alice Gandy, 2Dr Gavin Welsh, 1All Wales Medical Genomics Service 2Bristol University, Bristol*

Abstract - The Wilms' Tumour 1 (WT1) Gene on chromosome 11 is an enigmatic element of the human genome crucial to renal system and urogenital tract development. Mutations can cause malformation syndromes and can act as an oncogene in aggressive renal tumours. These have a poor prognosis and are common in infants, treated by long-term and often brutal chemotherapy and surgery. WT1 mutations vary dependent upon location; Denys-Drash and Frasier syndrome are more common and characterised by gonadal dysgenesis with a high risk of malignant tumours. There is currently no cure for these syndromes that are becoming more common and costly. Developing a gene therapy to target Denys-Drash syndrome (DDS) will fill a gap in the literature to prevent malformation on a genome level. Gene therapy has become more prevalent and the ability to transduce renal cells has been demonstrated with the specific expression of podocytes using Adeno associated viral (AAV) vectors. Clinical trials on T-cell receptor therapies have been successful in targeting WT1 antigens in leukaemia patients. In-utero administration can prevent infertility common in DDS due to gonadal dysgenesis and improve life quality. The use of AAVs and lipid nanoparticles are promising and will be compared in a subsequential part of this project where they will be trialled in murine models to ascertain the most systematic vector and administration method for a cost effective and safe large scale clinical trial at different life stages.

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Title - Postpartum Psychosis as a Genetically Distinct Manic Subtype: A Case-Control Analysis of Polygenic Risk

Authors - *[1]N. A. Khan [1]Cardiff University, Wales*

Abstract - Background: Postpartum Psychosis (PP) is a severe psychiatric emergency linked to Bipolar Disorder (BPD). While childbirth is a primary trigger, the specific genetic architecture, particularly the contributions of manic and depressive liabilities, remains poorly defined. This study aimed to quantify the genetic liability of PP and determine if it possesses a distinct profile compared to non-postpartum BPD.

Methods: 9,151 women were analysed: first-onset PP cases, women with BPD, and healthy controls. Polygenic risk scores (PRS) for BPD, Schizophrenia (SCZ), and Major Depressive Disorder (MDD) were generated via PRS-CS. Logistic regression, adjusted for ten principal components, examined: (1) PP cases vs. controls; and (2) PP cases vs. BPD. Variance was quantified via Nagelkerke, and multiple testing controlled via FDR.

Results: BPD PRS was a robust predictor of PP relative to controls (OR=2.24, 95% CI: 1.97–2.54, P<0.001), explaining 7.39% of variance. Crucially, compared to BPD, PP cases exhibited a significantly lower genetic load for MDD (OR=0.84, 95% CI: 0.74–0.96, FDR=0.015). No differences observed for BPD or SCZ liability (P>0.05).

Conclusions: While PP shares a substantial genetic substrate with BPD, it is characterized by a lower depressive genetic burden. This suggests PP represents a genetically "pure" manic or psychotic phenotype. Findings support the clinical distinction of PP and demonstrate the utility of PRS in dissecting Bipolar Disorder heterogeneity.

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Title - Reanalysis of genomic data doubles the diagnostic yield for Welsh patients recruited to the UK 100,000 Genomes Project

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Abstract - The 100,000 Genomes Project (100KGP) undertook genome sequencing of patients with rare diseases and cancer to study the role that genes play in disease, and to integrate genomics into UK healthcare. To contribute to 100KGP, the Wales Genomic Medicine Centre (a partnership between the NHS All Wales Medical Genomics Service (AWMGS), Cardiff University and Genomics England) recruited 438 individuals from 154 families that had been subject to pre-genomic genetic testing without reaching a diagnosis. The majority of probands (64%, 98/154) had neurological or neurodevelopmental phenotypes. Genome sequencing, variant calling, gene-based filtering and variant prioritisation were performed by Genomics England. AWMGS undertook clinical interpretation, validation and reporting of variants. Initial diagnostic yield was 20.8% (32/154) with variants of uncertain significance (VUS) reported for 11 more families. Most of the initial findings (81.4%, 35/43) could have been detected by clinical exome sequencing which was standard of care in Wales at the time. Reanalysis of the 100KGP Wales data using updated variant prioritisation tools, expanded gene lists, re-phenotyping and segregation studies, has increased diagnostic yield to 42.2% (65/154). RNA analysis was used to clarify the clinical significance of VUS in COQ4, ENPP, GATAD2B, NCAPD2, and THOC2. These findings demonstrate the clinical utility of genome sequencing, RNA analysis, and periodic reanalysis of genomic data.

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Title - Who Will PROM Go to Prom With? - Evaluating the SIDES Algorithm as a Machine Learning Approach for Patient Subgroup Identification in Breast Cancer Patient-Reported Outcome Measure Data

Authors - *Sarah Babiker, Cardiff University, Wales*

Abstract - Breast cancer patients can have very different experiences of treatment, yet standard analyses of Patient-Reported Outcome Measures (PROMs) often treat patients as a single group. This study applies the SIDES subgroup identification algorithm to a breast cancer PROM dataset to test whether it can identify meaningful patient subgroups based on quality-of-life outcomes. SIDES was originally developed for clinical trial settings, so this work explores how the method performs when applied to observational PROM data where trial structure is absent. The findings are used to assess where SIDES fits within the broader toolkit of machine learning approaches for subgroup identification in clinical research.

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Title - Switches and Signals: Mapping Phosphatase–EMT Signalling Associations in Breast Cancer Transcriptomes

Authors - *Jehosheba Tijani - Affiliated with: Esther Liu , Cardiff University*

Abstract - The epithelial-mesenchymal transition is a transcriptional mechanism associated with tumour invasion, metastasis and therapy resistance in breast cancer. Whilst the kinase signalling network involved in the EMT has been studied extensively, the role of protein phosphatases, which are key regulators of phosphorylation signalling, remains vague at the transcriptomic level. The objective of this project was to develop and implement a reproducible transcriptomics analysis pipeline, that identifies associations between phosphatase and EMT related gene programmes utilising RNA-seq data from the TCGA Breast Cancer (TCGA-BRCA) cohort. An R-based computational pipeline was designed with the express aim of analysing the TCGA-BRCA RNA-sequencing data. A curated list of human protein phosphates was used to profile phosphatase expression across the tumour samples. EMT pathway activity was quantified using single-sample Gene Set Enrichment Analysis (ssGSEA) with chosen EMT gene sets. Spearman correlation analysis was then performed to evaluate the interconnections between the phosphatase expression and EMT scores , with multiple testing correction applied using the Benjamini-Hochberg false discovery rate. Additionally, an exploratory correlation-based network was generated to better visualise the links.

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Title - Additional Data from the "Seasonal variation in the association between the rs2228145 variant of the IL-6R gene and self-reported long-COVID symptoms" clinical study.

Authors - *Katie Rees 1, Rebecca Aicheler 1, Lee Butcher 1, Keith Morris 1, Richard Webb 1, Isabel Massey 1, Ceri Lynch 2 , Lisa Roche 2, Alan Dodd 2, Brian Tennant 2, John Geen 2 1 - Department of Biomedical Sciences, Cardiff Metropolitan University, Cardiff CF5 2YB, Wales, UK 2 - Cwm Taf Morgannwg University Health Board, Royal Glamorgan Hospital, Llantrisant, Rhondda Cynon Taf CF72 8XR, Wales, UK*

Abstract – This poster presents data from the recent CTMUHB/Cardiff Met collaborative study "Seasonal variation in associations between the rs2228145 variant of the IL-6R gene and long-COVID symptoms". The study aimed to collect DNA and plasma samples and Long-COVID Symptom Questionnaire responses from 192 individuals with prior COVID-19 infection. Here, we present three unpublished study outcomes regarding participants' symptoms within specific long-COVID symptom-clusters, vaccination status/delivery modes, and blood-borne Vitamin D levels within the cohort.

Potential associations were evaluated between participants' rs2228145 genotypes, COVID-19 severity/frequency, or pre-existing conditions and: 1) Long-COVID symptom-cluster scores; 2) vaccination status/delivery mode; 3) Vitamin D levels, using univariate logistic regression and chi^2 analyses. Initial COVID-19 severity and pre-existing arterial hypertension were the strongest predictors of persistent long-COVID symptoms across most symptom clusters (eg. chronic fatigue, cognitive impairment, cardiopulmonary complications, psychological disorders). Repeated infections (≥3) were significantly associated with chronic fatigue and neurological symptoms, while prior vaccination against COVID-19 showed protective effects against several symptom clusters. Conversely, neither vaccine delivery mode, nor Vitamin D levels were associated with Long-COVID risk.

These results highlight the importance of initial disease severity, reinfection, and underlying health conditions as key Long-COVID risk factors, while highlighting the protective role of vaccination. These findings may potentially inform future Long-COVID management and treatment strategies.

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Title - Lipidomics and Metabolomics for Rare Disease Diagnosis

Authors - Ali Asgari, M., Griffiths, WJ., Wang, Y. Swansea University Medical School, Wales

Abstract – The diagnostic odyssey represents one of the major challenges to people suffering from a rare disease. In an attempt to shorten this journey, and thanks to funding from MRC and NIHR, we are utilising the new "omics" technologies of lipidomics and metabolomics to investigate patient plasma samples to help give them a diagnosis faster. Once a diagnosis is in place we are also exploiting this technology to monitor response to new therapies. We are working closely with clinicians and scientists at the University Hospital of Wales, Guy's and St Thomas', St Mary's Hospital Manchester, Sheffield Children's Hospital amongst many others. Our ultimate goal is to translate these "omic" methods from the university research lab to the NHS metabolic lab.

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Title - More than you can imagine: an anthology of rare experiences

Authors - Natalie Frankish1, Rachel McEleny1, Jennifer Barley1 – Genetic Alliance UK

Abstract - The anthology is a powerful collection of creative works that bring to life the experiences of individuals within the genetic, rare, and undiagnosed communities through poetry, personal stories, photography, and artwork.

Featuring over 60 submissions, the anthology represents more than 50 different rare conditions. Contributors range in age from 9 to 71 and come from across the UK, including individuals living with rare conditions, their families, parents, partners, siblings, children, healthcare professionals, and support organisations.

Themes of resilience, isolation, mental health, and the experience of 'fighting' for care echo throughout the anthology. But so too does the strength found in support and community. While each story is deeply personal, it is impossible to ignore the shared challenges. Delayed diagnoses, a lack of awareness among healthcare professionals, fragmented care, and barriers to accessing treatment and support.

Our goal with this anthology is to amplify the voices of those within the genetic, rare and undiagnosed community to achieve greater understanding of the impact of these conditions. We hope these powerful creative expressions will inform the public, healthcare professionals, policymakers, and parliamentarians and drive meaningful change.

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Title - Genomics Partnership Wales: Our approach to Patient and Public Engagement

Authors - *Genomics Partnership Wales*

Abstract – As genomic medicine becomes increasingly integrated into healthcare, engaging patients and the public is essential to ensure transparency, trust, and equitable access. Genomics Partnership Wales was created to ensure that a collaborative approach is taken in the development of genomics in Wales for the benefit of patients and the population.

GPW has two PPIE groups, the Sounding Board and the Genomics Ambassadors.

The Sounding Board is a PPIE group made up of people with lived experience of rare and genetic conditions and aims to help improve patient experiences and clinical services for better health and wellbeing of people living with rare and genetic conditions. Sounding Board members use their lived experience to influence organisations, workstreams and strategies for genomics in Wales.

Genomics Ambassadors is a PPIE group made up of past members of the Sounding Board, who use their lived experience and Sounding Board experience to influence peers, communities and the public to raise the profile of genomics in Wales. The ambition for this group is to retain expertise and insights of the Sounding Board member, support Delivery Plan deliverables relating to PPIE and co-production and raise the profile of genomics in Wales.