2026 Conference proceedings

WMGL
STP CONFERENCE

LONG READ SEQUENCING

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Programme:

WMRGL STP Conference

09:30 to 17:00 on 24/04/2026 at Edgbaston Park Hotel (53 Edgbaston Park Rd, Birmingham B15 2RS).

Session	Start	End
Welcome and sign in Complementary refreshments will be served to all guests. This will include Tea, Coffee and pastries. During this time, the guests will be allowed to mix and network	09:00	09:15
Overview of the day Host will welcome everyone and go though the agenda for the day.	09:15	09:30
Guest talk: Implementing long read sequencing in rare disease diagnostics. Speaker: Lorraine Hartles-Spencer	09:30	10:10
Guest talk: Development and Clinical Implementation of a Standardised Nanopore Bioinformatics Pipeline for Rare Disease Diagnostics. Speaker: Nour Mahfel	10:15	10:55
Mid-Morning break (Complementary tea, coffee, cereal bar and fruits served)	11:00	11:15
Guest talk: There's More to Nanopore. Speaker: Luke Ames	11:15	11:55
Lunch Break Hot and cold buffet served at the hotel restaurant	12:00	13:00
Pre-workshop seminar - Trust Social media policy Speaker: Speaker from the trust	13:00	13:30
Workshop STPs will group to discuss and reflect on the lectures. STPs split into groups to audit their digital footprint and complete the reflection document.	13:30	15:00
Selected oral presentations from abstracts		
Missing Heritability in Inherited Cancer Genes Speaker: Leanne Herber	15:00	15:20
Life on Ice: Comparing the impact of vapour freezing and slow programmable freezing on post-thaw semen parameters; vitality, motility and DNA fragmentation Speaker: Adele Morgan	15:20	15:40
Poster sessions 12 posters will be selected from the abstracts submitted. The guests will be encouraged to network. Complementary tea, coffee and a sweet treat served	15:40	17:00

Implementing long read sequencing in rare disease diagnostics

Speaker: Lorraine Hartles-Spencer.

Notes:

Development and Clinical Implementation of a Standardised Nanopore Bioinformatics Pipeline for Rare Disease Diagnostics

Speaker: Nour Mahfel

Notes:

There's More to Nanopore

Speaker: Luke Ames

Notes:

Pre-workshop seminar - Trust Social media policy

Speaker: Isobel Hunter

Notes:

Workshop

S-C1-0 TA-04 - Audit your digital footprint for adherence to your organisations policy and professional standards and make recommendations for future professional and personal practice.

Considerations:

Considerations

- Advantages and disadvantages of digital communication and social media
- Standards; local, national, and professional
- The professional and personal standards of a Clinical Scientist
- Exercising professional judgement and personal responsibility
- Ethical practice, including confidentiality, consent and candour
- Responsibility to share knowledge
- Sharing good practice
- The impact of understanding healthcare science on patients and public and engagement in their care
- Stakeholder and patient involvement
- Drivers and limitations
- Culture and values

Trainers: Eudmar Marcolino **One File Assessor:** Anita Luharia and Jennifer Whitfield

Pre-Training reflection	
How might improving your online presence positively impact patient care and public engagement?	
Where do you draw the line between personal and professional use of social media?	
What risks might arise if patient information (even indirectly) is shared online?	
How might tone or language online affect trust in healthcare professionals?	

What responsibilities do you have to protect public confidence in your profession through your digital presence?	
What benefits could auditing your digital footprint bring to your professional development?	

Digital Footprint Audit Template

Audit completed → Sections 1–6

Reflection → Section 7

Action plan → Section 8

Section 1: Identification of Platforms	
Which online platforms do you currently use (e.g., LinkedIn, Twitter/X, ResearchGate, Facebook, Instagram)?	
Are these accounts personal, professional, or mixed use?	
Do you separate professional and personal accounts where appropriate?	

Section 2: Professional Standards & Policy Alignment	
Professional profiles	
Does your profile clearly identify your professional role and affiliation?	
Are your posts consistent with organisational policy (e.g., confidentiality, candour, inclusivity)?	
Do you share content that could be misinterpreted or conflict with NHS values?	

Do you use professional judgement when deciding what to post or share?	
Personal Profiles	
Do you maintain appropriate boundaries with service users, carers, and colleagues online?	
Have you ever accepted friend requests or direct contact from service users on personal accounts?	

Section 3: Confidentiality & Patient Impact	
Have you ever shared patient-related information, even indirectly (e.g., case details, photos)?	
Could any content be perceived as breaching confidentiality or professional boundaries?	
How might patients or the public interpret your online presence?	
Do you understand that even indirect references may breach confidentiality?	

Section 4: Communication & Tone	
Is your communication always polite, respectful, and inclusive?	
Do you avoid discriminatory or offensive material?	
Are you transparent about your role when sharing knowledge or opinions?	
When engaging in debate, are your views evidence-based and accurate?	

Section 5: Honesty & Trustworthiness	
Does your online conduct uphold public trust and confidence in your profession?	
Do you check the accuracy of information before posting or endorsing it?	
Do you make clear when views are personal (e.g., disclaimers)?	

Section 6: Risks & Opportunities	
What are the potential risks of your current digital footprint (e.g., reputational, legal, ethical)?	
What opportunities exist (e.g., sharing best practice, public engagement, professional networking)?	
Do you follow your employer's social media policy?	
Personal Profiles	
Do you use privacy settings appropriately to manage personal vs professional boundaries?	

Section 7: Reflection	
What strengths did you identify in your digital footprint?	
What areas need improvement in light of HCPC standards?	
How does your digital presence align with NHS values and HCPC standards of conduct, performance, and ethics?	

Section 8: Action Plan	
What immediate changes will you make (e.g., update	

bio, adjust privacy settings, remove content)?	
What longer-term commitments will you adopt (e.g., regular audits, professional content sharing)?	
How will you monitor progress and ensure accountability?	

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A Service Evaluation of Paediatric and Syndromic Cardiomyopathy Genetic Testing at Oxford Regional Genetics Laboratories

Violet Simpson[1]

[1]Faculty of Biology, Medicine and Health, The University of Manchester, Manchester, England; Oxford University Hospitals NHS Foundation Trust, Oxford, England.

Introduction: Cardiomyopathies are myocardial disorders associated with heart failure and sudden cardiac death, for which genetic testing is crucial to clinical management. This study evaluated R135 paediatric and syndromic cardiomyopathy testing at OUH, where WGS has been implemented since 2021 alongside singleton WES-based large gene panel testing for urgent cases; this service has not been evaluated since implementation, highlighting the need for assessment, which is further exacerbated by the context of the rarity, severity, and heterogeneity of childhood-onset disease.

Methods: A retrospective analysis of all samples referred for R135 testing at OUH between October 2019 and September 2025 was performed.

Results: A total of 134 probands were included, the majority aged ≤20 years (83.6%). The most common phenotypes were HCM (n = 49, 36.6%) and DCM (n = 44, 32.8%), followed by unspecified cardiomyopathy (n = 17, 12.7%), other phenotypes (n = 12, 9.0%), and RCM and LVNC (n = 6, 4.5% each). Referrals were received from 29 centres across five specialties.

Overall, a genetic diagnosis was identified in 32 cases (23.9%), while 80 (59.7%) had no identifiable genetic cause and 19 (14.2%) yielded VUS. Diagnostic yield differed significantly by testing modality and age, with higher rates of genetic VUS observed in WES compared with WGS ($p < 0.05$), as well as higher yield in individuals aged >20 years ($p = 0.016$).

In the WGS cohort (n= 58), five cases received a diagnosis through gene-agnostic analysis, including variants identified via *de novo* analysis, Exomiser prioritisation, and additional panel application; these would have been missed using the semi-urgent WES pathway.

WES demonstrated shorter turnaround times compared with WGS; suggesting WES remains preferable in clinical scenarios where rapid results are required. However, the margin between testing approaches appears to be decreasing. Though the extent of this is uncertain with majority of data relating to earlier stages of the pathway not being available.

Conclusions: This study provides a real-world evaluation of R135 genetic testing at OUH and establishes a standard for future audit and optimisation. Further conclusions are limited by the small sample size, cohort heterogeneity and changes in laboratory practice over time.

Keywords: paediatric and syndromic cardiomyopathy, service evaluation, gene agnostic WGS

An Evaluation of Genomic Visualisation Tools to support the Clinical Classification of Copy Number Variants, Structural Variants and Uniparental Disomy within Prenatal WGS cases

Kavi Jeshram[1]

[1] Clinical Genetics Department, Great Ormond Street Hospital

The increasing usage of whole genome sequencing (WGS) in prenatal diagnostics has significantly improved the detection of genomic variation, specifically copy number variants (CNVs), structural variants (SVs) and uniparental disomy (UPDs). This enhanced diagnostic yield contributes to an increase in equitable healthcare as more patients with complex genomic mechanisms can be diagnosed. More expecting mothers can then make informed pregnancy and family planning choices. However, both the volume and complexity of the data have increased, potentially impacting turnaround times. To mitigate delays in reporting, visualisation tools can be implemented to enable parallel viewing of; detailed supplementary information for classification such as database information, and detailed variant information such as breakpoint locations, which enhance classification speed. Despite the availability of multiple genomic visualisation tools, few have been evaluated for their suitability within clinical classification workflows. This study aims to recommend a visualisation tool that could support clinical scientists with these workflows, specifically within prenatal diagnostics. Tool requirements, derived from a questionnaire distributed to clinical scientists and a literature review, informed the creation of both a preliminary and a secondary scoring rubric. Another literature search was then conducted to identify candidate visualisation tools, with the initial scoring rubric used to organise development priority. A mixed agile methodology was used to test these tools, using previously hard to classify variants as a test set. Each tested tool was evaluated using the secondary scoring rubric, focusing on functionality, usability, integration capability, and support for genomic annotations. The highest-ranked tools were presented to stakeholders for user testing, during which usability issues and scores were recorded. Final scores were then calculated. Among the evaluated tools, JBrowse2 achieved the highest overall score. It provided interactive exploration of genomic regions, effective visualisation of large variants, clear representation of genes and transcripts, and the ability to integrate multiple annotation sources. In contrast, other tools such as Samplot and SVHawkeye offered more limited functionality. Overall, JBrowse2 provides the best combination of usability, scalability, and integration potential. The study therefore recommends JBrowse2 as a suitable visualisation platform to support clinical interpretation of genomic variants in prenatal WGS workflows.

Keywords: genome browser, visualisation tools, prenatal variants

Development of a New SMN1 Deletion Testing Protocol to Improve SMA Service

Olivia Iyayi[1]

[1]West Midlands Genetics Laboratory, Birmingham Women's and Children's NHS Foundation Trust, Birmingham

Spinal Muscular Atrophy (SMA) is an autosomal recessive genetic disorder affecting approximately 1 in 10,000-20,000 live births worldwide. It is caused by biallelic loss-of-function variants in the *survival motor neuron 1 (SMN1)* gene, which leads to progressive degeneration of motor neurons and muscle atrophy. Approximately 60% of affected individuals present with a severe form of SMA, which, if untreated, can be fatal within the first two years of life. Available treatments can halt further neuronal loss by preserving motor neurons, but they cannot replace neurons that have already degraded. Therefore, early diagnosis is critical to improve patient prognosis, as shown in preclinical and clinical studies.

Diagnostic testing is performed using Multiplex Ligation-dependent Probe Amplification (MLPA) at this Genomic Laboratory Hub (GLH). Although SMA samples are prioritized over other urgent cases, the MLPA procedure is time-consuming. Results are available after approximately three days under optimal conditions. An audit performed at this GLH found that the turnaround time (TAT) for MLPA testing in neonatal patients referred for SMA testing was 12 days in 2025. According to the National Institute for Health and Care Excellence (NICE) guidelines, treatment can only begin following confirmation of *SMN1* deletion and determination of *survival motor neuron 2 (SMN2)* gene copy number, which is used to predict disease severity and treatment eligibility. This further emphasises the importance of minimising the time between sample receipt and reporting results. The current TAT may limit the ability to deliver rapid results for urgent neonatal patients.

Introducing a more rapid diagnostic test for SMA is essential to facilitate earlier initiation of treatment and improve clinical outcomes for these patients. While other laboratories have adopted new testing methods to meet the demand for rapid SMA diagnosis, no such method is currently available within this GLH. The overall aim is to validate and verify a new rapid SMA testing method for this GLH, which can detect both *SMN1* and *SMN2* copy number, thereby improving clinical service delivery.

Keywords: Spinal Muscular Atrophy (SMA); *survival motor neuron 1 (SMN1)* gene; copy number detection.

Examining the Clinical Utility of Long Read Sequencing in the Paediatric Rare Disease Setting

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Roughly 7000 rare diseases have been identified to affect approximately 300 million people globally. Of these, 80% are predicted to have an underlying genetic contributor, and over half have early onset within childhood. Despite the emergence of whole genome sequencing (WGS) technologies, many patients still present with clinical features indicative of a rare disease, but a genetic basis is not found. A diagnosis can focus clinical management for the patient, and help identify recurrence risk for the family.

Short-read WGS has been instrumental in identifying novel single nucleotide variants (SNV), small indels, and copy number variants (CNV) associated with many rare diseases. Its utility is reflected by the presence of WGS panels for many indications in the National Genomic Test Directory, for both rare disease and cancer. This utility is further exemplified through its ability to provide diagnoses to ~25% of patients who had previously exhausted all standard of care testing. However, the question remains: what could be causing disease in the remaining 75%? This could be because: the cause is not genetic, short-read WGS cannot detect the relevant change, or a variant lies outside the reviewed regions (particularly if analysis encompasses only exonic regions or is not gene-agnostic).

Long-read sequencing (LRS) uses primarily Pacific Biosciences (PacBio) and Oxford Nanopore (ONT) platforms, with a reported 7-17% increase in rare disease diagnostic yield. Unlike short-read, LRS generates reads, which may span megabases, better suited to detect structural variants (SVs) such as inversions and translocations. LRS reads also allows for increased coverage of regions with high sequence similarity, phasing of variants, epigenetic and transcriptomic analyses, and faithful sizing of repeat expansion regions. Therefore, LRS technologies represent a promising solution for patients who still lack a genetic diagnosis following short-read WGS.

This project is in collaboration with the NIHR BioResource study, 100 families without genetic diagnoses will be sequenced via ONT, and analysis will be carried out by the West Midlands genetics laboratory alongside the NIHR research teams. We hope that the outcomes of this project will support the implementation of LRS within paediatric rare disease diagnostics.

Keywords: Long-read sequencing, diagnostic yield, clinical utility

Life on Ice: Comparing the impact of vapour freezing and slow programmable freezing on post-thaw semen parameters; vitality, motility and DNA fragmentation

(Selected for oral presentation)

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Sperm is cryopreserved when a patient requires treatment that could negatively impact their fertility (e.g. chemotherapy). For some this may be their only hope of having biological children. However, even optimal freezing techniques create mechanical and chemical stress that can cause cryoinjuries. 25-75% of sperm can be rendered non-viable or immotile following cryopreservation. Cryopreservation can also induce sublethal damage, such as DNA fragmentation, which has been linked with decreased fertilisation rates, increased risk of miscarriage and poorer fertility treatment outcomes. However, despite much research there is no field consensus on which freezing method preserves the greatest yield of viable motile sperm with intact DNA .

Birmingham Women's Fertility Centre uses vapour freezing (VF), where semen is incrementally lowered by hand into liquid nitrogen over 35 minutes. In contrast, slow programmable freezing (SPF) uses a machine to cool the sample gradually over 2-4hrs. The majority of cryoinjuries are caused by inappropriate freezing rates, therefore, it is hypothesised that the more controlled SPF will have a positive effect on post-thaw semen parameters. This pilot study will compare the impact of VF and SPF on sperm motility and DNA integrity. Semen samples will be assessed pre and post VF and SPF. Motility will be assessed using Flagellar Analysis and Sperm Tracking (FAST) and DNA fragmentation will be assessed using the TUNEL assay. Statistical analysis, using Mann Whitney U, will determine if SPF has a positive effect on post-thaw semen parameters in comparison to the department's current VF method.

Oncology patients can be heavily impacted by cryoinjury as they can have increased DNA damage levels and patients with poorer initial semen parameters experience a greater loss of sperm quality after freezing. In addition, the short diagnosis-to-treatment window means they have reduced opportunities to preserve samples. Since patients rely on sperm cryopreservation to preserve their fertility it is imperative that freezing methods minimise the rate of cryoinjury and optimise post-thaw survival. This project aims to assess if the introduction of SPF to clinic practice could improve the yield of high quality sperm for groups impacted by cryoinjury, such as oncology patients.

Keywords: Sperm Cryopreservation, vapour freezing, slow programmable freezing

Missing Heritability in Inherited Cancer Genes

(Selected for oral presentation)

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Familial adenomatous polyposis (FAP) is an autosomal dominant inherited condition caused by loss of the tumour suppressor gene adenomatous polyposis coli (APC), leading to the development of numerous colonic polyps and a near 100% lifetime risk of colorectal cancer if untreated. Currently, a subset of patients represent a diagnostic odyssey, exhibiting clear phenotypes associated with FAP with no identifiable likely pathogenic/pathogenic variants using current sequencing methods. A previous in-house study utilised Oxford Nanopore Technology (ONT) to perform whole genome sequencing where a deeply intronic APC variant was identified in a patient affected with FAP.

Here, we present a proof-of-principle study using a long-range PCR combined with ONT sequencing approach to capture the entire APC gene. Primers used in the long-range PCR were designed to amplify overlapping 10–12 kb fragments covering the full APC gene. PCR conditions were optimised on control genomic DNA until amplicons of expected sizes were obtained. Sequencing of these amplicons confirmed alignment to the APC gene, demonstrating that the approach is able to capture the correct genomic regions. This work establishes a potential method for comprehensive sequencing of the APC gene. Further work is needed to assess the feasibility of this approach to detect variants using patient samples. This project may provide patients with access to further testing which may confirm a diagnosis and aid with cascade testing.

Keywords: missing heritability, sequencing, colorectal cancer

Preparing NHS Genomics clinical scientists for ethical decision-making: a mixed-methods study of trainee and alumni perspectives on ethics teaching within the NHS (Genomics) Scientist Training Programme (STP)

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[1]Synnovis Analytics

The ethical dimension of the work of National Health Service (NHS) genomics clinical scientists is growing as genomic medicine becomes increasingly integrated into routine healthcare. This growth is driven by advances in genomic technologies, the incorporation of genetic/genomic testing into mainstream healthcare services, and political initiatives such as Genome UK. Clinical scientists are encountering complex ethical issues, including the management of incidental findings and predictive testing in children, with increasing frequency.

However, little is known about whether current training is sufficient to prepare clinical scientists to navigate such ethical issues. This study aimed to explore the perceptions of ethics teaching within the genomics Scientist Training Programme (STP) in relation to preparing trainees for ethical decision-making in practice.

A mixed-methods approach combining Likert scale survey items and free-text items was conducted with current genomics STP trainees (n=39) and STP-trained genomics clinical scientists (n=18). Likert scale data were analysed descriptively, and qualitative data were analysed using reflexive thematic analysis, and findings integrated through convergent design.

Findings from both cohorts suggest that current STP ethics teaching is widely perceived as overly-theoretical and insufficiently aligned to genomics clinical scientist practice. Workplace experiential learning was identified as the most valuable mode of ethics teaching, being reported in association with confidence when approaching ethical decision-making. However, access to such opportunities was reported to be variable. Alumni reported greater dissatisfaction with current ethics teaching than current STP trainees, suggesting that deficits become more apparent in independent practice. Consistency in qualitative themes across cohorts suggests a persistent perception of current ethics teaching in the STP as lacking, with a strong desire for practical scenario-based ethics teaching in both university and workplace settings.

Study findings suggest that current approaches to ethics teaching within the STP may be insufficient to cultivate consistent ethical reasoning and decision-making skills in clinical scientists. Recommendations for teaching amendments, such as mandatory workplace ethics training activities and scenario-based university teaching, are discussed.

Keywords: ethical preparedness, Scientist Training Programme, ethics education

Beckwith-Wiedemann Syndrome and Silver-Russell Syndrome Testing – Breadth or Depth?

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Beckwith-Wiedemann syndrome (BWS) and Silver-Russell syndrome (SRS) are imprinting disorders, characterised by overgrowth and growth restriction respectively. Both conditions are genetically heterogeneous, involving a range of genetic and epigenetic mechanisms such as aberrant methylation, UPD, CNVs and SNVs (Õunap, 2016). Diagnosis is further complicated by mosaicism and tissue-specific methylation differences (Schlaich, Hubens and Eggermann, 2023).

In some patients, aberrant methylation occurs at multiple imprinted loci across the genome, termed multi-locus imprinting disorder (MLID). These individuals may show a predominant BWS or SRS phenotype or have a blended presentation with features associated with multiple imprinting disorders (Eggermann *et al*., 2016). These individuals are often referred for BWS/SRS diagnostic testing in the first instance.

Current MS-MLPA frontline testing can detect CNVs and methylation changes at 11p15 (ME030) (as well as Chr7 and Chr14 for SRS referrals), followed up with testing at multiple imprinted loci (ME034) when MLID is suspected. However, these assays have limited sensitivity for low-level mosaicism and cannot detect all variants associated with BWS, SRS or MLID (Schlaich, Hubens and Eggermann, 2023).

As a result, many patients referred for BWS/ SRS testing have a long diagnostic odyssey, often requiring multiple tests on different tissue types. There is currently a gap for an assay capable of detecting a wider range of molecular changes with higher sensitivity to detect low-level mosaicism.

This project will aim to look at five years' worth of data from patients tested for BWS and SRS at the West Midlands Genetics Laboratory (WMRGL). By analysing this information, we can determine the average number of tests per patient, overall costs and time taken to reach a diagnosis. We will also review emerging technologies, such as ddPCR, ASMM RTQ-PCR and methylation sequencing panels to determine if they can offer improvements in coverage and/ or mosaicism detection, helping to improve diagnostic efficiency and consequently patient outcomes. The overall aim of this project is to develop an improved testing pathway for BWS and SRS that gives patients quicker results and reduces unnecessary testing, making best use of NHS resources and ultimately improving care for affected children and families.

Validation of Avenio Edge and RMHv3 Solid Cancer NGS Panel

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Recent validations at the WMGL have focused on automating and expanding the solid cancer service. The Roche Avenio Edge performs automated NGS library preparation and target enrichment, reducing staff hands on time whilst minimising variability. It is optimised for FFPE-derived DNA, enabling the creation of enriched libraries ideal for solid tumour analysis. The RMHv3 custom panel, designed by the Royal Marsden, consists of 250+ genes in line with the genomic test directory and was selected to offer the benefit of large panel testing to more cancer patients.

The aim of the validation was to ensure the Avenio Edge could produce enriched libraries adequate for NGS analysis and to validate the RMH panel to deliver on sequence variants and copy number variants in order to maintain current and future test directory compliance.

Validation was conducted using clinical solid tumour samples and EQA cases previously tested via orthogonal methods, alongside commercial controls. Key validation criteria included concordance, precision, repeatability, and robustness, and sought to determine essential parameters of the assay.

The automated workflow has demonstrated improved output and minimal hands-on time, whilst maintaining consistent library quality and sequencing coverage metrics. The RMH panel has shown high levels of concordance with orthogonal methods and metrics optimal for routine clinical use within solid cancer workflows, including low minimum DNA input and low LoD to detect low-level genetic variants.

This validation demonstrates the clinical utility of the change to deliver high quality NGS analysis and supports enhanced efficiency and standardisation of the solid cancer workflow.