Summer Scientific Meeting 2024
10th – 11th June 2024, ICC, Birmingham

Day 1 Programme

09:00-09:45	***Registration/Coffee/Posters***	
09:45	Introduction to meeting	
Session chair:		
09:50-10:20	Title TBC	Simon Paine
10:20-10:35	Long Read Whole Genome Sequencing for Central Nervous System Tumours with Oxford Nanopore Sequencing	Dr Rowan Howell, Genomics England
10:35-10:50	Nanopore long-read sequencing as a supplementary technology for short-read whole genome sequencing	Joseph Norman, South West GLH, Bristol
10:50-11:05	The use of long-read Nanopore sequencing with adaptive sampling to detect and characterise pathogenic genomic structural variations in patients with myeloid neoplasms	Matthew Salmon, Central & South GLH, Wessex
11:05-11:15	title tbc (Illumina)	Greg Elgar, Genomics England
11:15-11:45	***Coffee/Trade/Posters***	
Session chair:		
11:45-12:00	*PTPMT1*: Shedding light on a new disease-causing gene with the NHS Highly Specialised Services (HSSs)	Padraig Flannery, Queen Square Centre for Neuromuscular disease, London
12:00-12:15	Preimplantation testing for mitochondrial DNA disease; determining the risk of transmission of serious mitochondrial disease	Kate Hickman, North East & Yorkshire GLH, Newcastle
12:15-13:00	***Lightning Presentation Session***	
1	Genetic diagnosis of FSHD – an increasingly complex picture	Melissa Ward, South West GLH, Bristol
2	An audit of rapid whole exome sequencing results for patients with suspected likely inborn errors of metabolism	Clare Clayton, Central & South GLH, Birmingham
3	Proposal to Investigate the Utility of Long-read Sequencing for the Diagnosis of Neurodegenerative Disorders	Eleanor Minshall, SE Scotland Genetic Service
4	DECIPHER – Empowering rare disease diagnosis and research through the sharing of phenotype-linked candidate diagnostic variants	Julia Foreman, DECIPHER, Cambridge
5	Applying updated ACGS Variant Classification Guidelines to Historic cases of Rare Disease, A New	Duncan Baker & Adam Hodgson, Julia Garnham Centre, Sheffield

	Opportunity for NHS HE Partnership, The Julia Garnham Centre	
6	A deep dive into phenylketonuria (PKU): Deep intronic variants activate pseudoexons in *PAH* and account for missing heritability	Rachael McNeilly, South West GLH, Bristol
7	Review of the first year of testing for hereditary alpha-tryptasemia (HAT)	Emma Miles, North West GLH, Manchester
8	PyGVP: a novel bioinformatics filtering package	Ashley Pritchard, North Thames GLH, London
9	Scaling clinical bioinformatics processes in the cloud	Adriana Toutoudaki, East GLH, Cambridge
13:00-14:00	***Lunch/Trade/Posters***	
Session chair:		
14:00-14:30	Living with a rare genetic condition: the patient and family perspective	Sarah Wynn, UNIQUE
14:30-15:00	Title TBC	Rob Taylor, North East & Yorkshire GLH, Newcastle; Sian Ellard and Emma Baple, South West GLH, Exeter
15:00-15:30	ACGS AGM	
15:30 – 16:00	***Coffee/Trade/Posters***	
Session chair:		
16:00-16:20	GenQA – EQA for the end to end genomic pathway	Professor Sandi Deans, Director GENQA
16:20-16:40	title tbc	Dr Rosa Morra, Scientific Program Manager, EMQN
16:40-16:50	title tbc	
16:50-17:00	title tbc	
17:00-17:10	title tbc	
17:15-18:45	**Drinks Reception @ the ICC**	

Association for Clinical Genomic Science

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Day 2 Programme

08:30-09:10	***Registration/Coffee/Trade/Posters***	
09:10	Introduction to meeting	
Session chair:		
09:15-09:30	Automating PVS1 classification for variant interpretation	Kieron Millard, All Wales Genomics Laboratory
09:30-09:45	The PS4 likelihood ratio calculator: enabling accurate and flexible allocation of evidence weighting for case control data	Charlie Rowlands, Institute of Cancer Research, London
09:45-10:00	Functional studies prove vital for reclassifying variants of uncertain significance (VUS). A highly specialised and rare disease diagnostic service perspective.	Charlotte Alston, North East & Yorkshire GLH, Newcastle
10:00-10:10	Title TBC	Alex Hobbs, Account Manager South, OGT
10:10-10:20	An AI driven platform to support the diagnostic process for germline and somatic applications	Michael Blum, SeqOne Genomics
10:320-10:30	Improving the BCR:ABL1 detection and monitoring with TRUdPCR	TBC
10:30-10:40	Evaluation of Archer VariantPlex: A target next generation sequencing panel for SNV and CNV detection in FFPE	Sophia Dixon, Central & South GLH, Birmingham
10:40-11:10	***Coffee/Trade/Posters***	
Session chair:		
11:10-11:30	Pharmacogenetics implementation in the NHS	Jessica Keen, Pharmacy Lead, North West Genomic Medicine Service Alliance
11:30-11:50	Title TBC	George Batchelor,
11:50-12:05	A New Genomic Instability Test for Ovarian Cancer	Joe Shaw & Katherine Sadler, North West Genomics Lab, Manchester
12:05-12:20	Classification of reduced penetrance variant in high penetrance genes: development of CanVIG-UK recommendations for cancer susceptibility genes	Alice Garrett, Institute of Cancer Research, London
12:25-13:10	***Lightning Presentation Session***	
10	A comprehensive RNA NGS fusion panel to detect actionable gene fusions for patients with solid tumours.	Robert Sansom, South West GLH, Bristol
11	Recommendations for laboratory workflow that better support centralised amalgamation of	Sophie Allen, Institute of Cancer Research, London

	genomic variant data: findings from CanVIG-UK national molecular laboratory survey	
12	Bespoke assay for confirmation of copy number variants in the pseudogene region of *PMS2*	Shona Borland, North East & Yorkshire GLH, Sheffield
13	Challenges and success of interpreting Whole Genome Sequencing (WGS) data for clinical care of patients with Acute Lymphoblastic Leukaemia (ALL)	Alona Sosinsky, Genomics England
14	Validation and Verification of the MethylationEPIC Array for classification of neurological tumours	Faryaal Safdar, Central & South GLH, Birmingham
15	Review of Pancancer fusion analysis for the sarcoma service at RMH NT-GLH	S MacMahon, North Thames GLH, London
16	R359 Service evaluation study on the first sequential 100 samples processed by NWGLH	Simina Botosneanu, North West GLH, Manchester
17	Breakpoint mapping using long read sequencing: a crucial step for classifying an *MSH2* exon 1-6 duplication	Paul Parsons, North East & Yorkshire GLH, Sheffield
18	The SINEs were there all along: Identification of a germline *DICER1* structural variant in a suspected *DICER1* syndrome case	Oluwatosin Taiwo, North Thames GLH, London
13:10-14:10	***Lunch/Trade/Posters***	
Session chair:		
	Best Practice guidelines	
14:10-14:25	EMQN best practice guidelines for genetic testing in hereditary breast and ovarian cancer	Miranda Durkie, North East & Yorkshire GLH, Sheffield
14:25-15:40	title TBC	Chris Wragg, South West GLH, Bristol
	title TBC	Miranda Durkie, North East & Yorkshire GLH, Sheffield & Emma-Jane Cassidy, Central & South GLH, Salisbury
	RD Cytogenomics Best Practice Guidelines Working Group: scope and progress to date	Mark Bateman, Central & South GLH, Salisbury & Deborah Morrogh, North Thames GLH, London
15:40-16:00	***Prizes, Closing Remarks and End of meeting***	

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Posters will be displayed in the foyer outside Hall 5/8. All authors must stand by their poster between 13:30-14:00 on Monday 10th June for questions.

Poster 1	Optimising AML patient care: Exploring an alternative sequencing method for accelerated identification of *FLT3*-TKD mutations.	Kerri Sweeney, West of Scotland Centre for Genomic Medicine
Poster 2	BCL6 rearrangements detected by FISH in Diffuse large B cell lymphoma (DLBCL) along with MYC and BCL2 rearrangements	Sajayan Joseph, LabPlus, New Zealand
Poster 3	NOT PRESENTING	
Poster 4	A consolidated, high through-put, testing service for myeloproliferative neoplasms used across North East & Yorkshire GLH	Dan Lock, NE & Yorkshire GLH, Leeds
Poster 5	Somatic TP53 mutations (sTP53m) in cell-free circulating DNA (cfDNA) in high-grade serous ovarian cancer (HGSOC)	Catherine Ramsden, North West Genomics Lab, Manchester
Poster 6	The North East & Yorkshire GLH pathway for managing germline cancer predisposition variants identified through haematological malignancy testing.	Philip Dean, NE & Yorkshire GLH, Leeds
Poster 7	PrimerDB: Platform for the efficient storage and management of primers	Mamoona Mushtaq, North West Genomics Lab, Manchester
Poster 8	The Julia Garnham Centre: Development and implementation of a variant classification digital learning environment, via staff student partnership	Maisy Wilkes, Julia Garnham Centre, London
Poster 9	Validation of an in-house HRD testing method	Lucas Pavlou, South East GLH, London
Poster 10	Shifting the paradigm of bioinformatics data storage: moving beyond CRAM and VCF files	Patrick Lombard, North Thames GLH, London
Poster 11	Beyond BED files - Developing a novel package to automate virtual gene panel filtering for clinical WGS	Sophia Johnson, North Thames GLH, London
Poster 12	CIP-API integration: Standardising the analysis of WGS and non-WGS variant interpretation at the North Thames Genomic Laboratory Hub	Maria Lock, North Thames GLH, London
Poster 13	Epigenomic meta-analysis of DNA from pre and post bariatric surgery patients.	Agnieszka Wisniewska, University of Derby,
Poster 14	*POLG* gene variant interpretation: comparative analysis of guidelines	Carl Fratter, Central & South GLH, Oxford
Poster 15	Improving the detection of small copy number variants in the rapid whole genome sequencing service for acutely unwell children	Suzanne Hocking, South West GLH, Exeter
Poster 16	Implementation of the Pregnancy Related Rapid Sequencing (PRRS) Service in NHS Wales	Fiona Kerr, All Wales Genomics Laboratory

Poster 17	The natural history of Classical Ehlers Danlos Syndrome	Chloe Angwin, London North West University Health Care NHS Trust
Poster 18	Prenatal Exome Sequencing for pregnancies with Fetal Anomalies – Reflection of 3 years of testing	Natalie Bibb, Central & South GLH, Birmingham
Poster 19	Gene burden tests in real-world patient cohorts – a new paradigm for NHS-led genomic discoveries?	Jade Doughty, East GLH, Cambridge
Poster 20	CNV reanalysis of exome data: Improving the diagnostic yield for rare diseases	Isabel Reid, Central & South GLH, Birmingham
Poster 21	Reducing the variant analysis burden in rare disease testing through semi-automated ACMG analysis	Joseph Halstead, All Wales Genomics Laboratory
Poster 22	Variant Reclassification in Expanded Carrier Screening: Implications for Personalized Management and Adaptation among Reproductive Couples	Yu Zheng, The Chinese University of Hong Kong
Poster 23	An evaluation of the Non-Invasive Prenatal Testing (NIPT) service in Wales for pregnancies where there are fetal anomalies on ultrasound scan	Sophie Bannister, All Wales Genomics Laboratory
Poster 24	Evaluating clinical presentation of *HNF1B* variants in a seven-year retrospective cohort	Erin Middleton, East of Scotland Regional Genetics Service
Poster 25	The pathogenic *PRRT2* variant evading tiered detection by whole genome sequencing	Helen Lord, Central & South GLH, Oxford
Poster 26	*STRC*-related hearing loss – insights from a large national diagnostic cohort	Molly Godfrey, North West Genomics Lab, Liverpool
Poster 27	Gene2Phenotype: high quality, detailed gene disease associations	Elena Cibrián Uhalte, European Bioinformatics Institute, Cambridge
Poster 28	From 'likely benign' to 'pathogenic': collaborative re-evaluation of an *IARS2* variant supports pathogenicity	Jack Baines, North East & Yorkshire GLH, Newcastle
Poster 29	Implementation and delivery of the R246 service (carrier screening at population risk for partners of known carriers) at the North West Genomics Laboratory Hub	Luke Stuart, North West Genomics Lab, Manchester