Summer Scientific Meeting 2022
11th – 12th July 2022, ICC, Birmingham

Day 1 Programme

09:00-10:00	***Registration/Coffee/Posters***	
10:00	Introduction to meeting	
Session chair:		
10:10-10:30	Genes and Health Program	Dr Shwetha Ramachandrappa
10:30-11:00	Evolution of MSI testing and clinical applications	Professor Sir John Burn, Translational and Clinical Research Institute, Faculty of Medical Sciences, Newcastle Upon Tyne Dr Ciaron McAnulty, The Newcastle Upon Tyne Hospitals NHS Foundation Trust
11:00-11:30	***Coffee/Trade/Posters*** **plus one 15 minute talk from CliniSys**	
11:15-11:30	GLIMS Genomics: Implementation of a bespoke flexible LIMS solution to the Regional Molecular Diagnostic Service, Belfast, Northern Ireland	Emma Huntridge, Genomics Business Development Director, CliniSys
Session chair:		
11:30-11:45	Assessment of the potential of a pre-emptive pharmacogenetic test to aid optimised drug treatment in the use of drugs associated with depression, anxiety and other mental health conditions	Jessica Woodley, West Midlands Regional Genetics Laboratory
11:45-12:00	Assessing the clinical utility of protein structural analysis in genomic variant classification: experiences from a diagnostic laboratory	Richard Caswell, Exeter Genomics Laboratory
12:00-12:15	A journey of variant interpretation from 'Tepid Class 3' to 'Class 5': The case of bi-allelic LoF variants in *NRROS*	Tariku Tadiso, St George's Genomics Service
	Lightning Presentation Session (8 total)	
12:20-12:25	DECIPHER – Introducing the new cardiac module	Julia Foreman, DECIPHER
12:25-12:30	Why do you do that? Quality Improvement in the South West Genomic Laboratory Hub (SWGLH)	Melanie Little, Bristol Genetics Laboratory
12:30-12:35	VariankBank: A flexible database and web application for the clinical analysis of variants from WGS and WES assays	Joseph Halstead, All Wales Medical Genomics Service
12:35-12:40	Utility of gene agnostic analysis in WGS and impact on improving analysis pipelines	Beatriz Diez Dacal, Oxford Regional Genetics Laboratories

12:40-12:45	Diagnoses Achieved Through Prenatal Exome Sequencing	Stephanie Allen, West Midlands Regional Genetics Laboratory
12:45-12:50	NIPT screening in Scotland – an 18 month review	East of Scotland Regional Genetics Service
12:50-12:55	The power of the gene-agnostic approach: R14 rapid trio WES identifies biallelic variants in a newly established disease gene causing neonatal cardiomyopathy	Hayley Lees, Exeter Genomics Laboratory
13:00-14:00	***Lunch/Trade/Posters* plus two 15 minute talks from Nonacus and Bionano**	
13:30-13:45	Cell free DNA molecular profiling in paediatric cancer patients	Reda Stankunaite, Nonacus
13:45-14:00	Next-Generation Cytogenomics with Optical Genome Mapping (OGM)	Amy Tibbo, Business Support Scientist, EMEA, Bionano
Session chair:		
14:00-14:20	Long Reads whole genome sequencing for Scottish families with rare disease	Dr Prasun Dutta, Research Fellow (Bioinformatician) Genome Medicine Group, University of Edinburgh
14:20-14:40	Complexities in the generation of genomics proficiency testing material accurately mimicking human specimens	Rachel Taylor, Scientific Programme Manager, EMQN
14:40-15:00	International System for Human Cytogenomic nomenclature (ISCN) – the challenge of the nomenclature	Melody Tabiner, Deputy Scheme Director (Cytogenetics), GENQA
15:00-15:30	ACGS AGM	
15:30-16:00	***Coffee/Trade/Posters* plus one 15 minute talk from Promega**	
15:45-16:00	Experience running OncoMate for clinical service (official title TBC)	Natalie Jorgensen, Senior Genetic Technologist, Manchester University NHS Foundation Trust
Session chair:		
16:00-16:15	Findings of the ACGS Workforce Development Committee Pre-Registration Scientist Survey 2021	Amy Webb, Wessex Regional Genetics Laboratory
16:15-16:30	Experiences of implementing an exome based nuclear gene panel test for diagnosis of mitochondrial disease	Conrad Smith, Oxford Regional Genetics Laboratories
16:30-16:45	UK Best Practice Guidelines for Genetic Testing for Mitochondrial Disease	Jack Baines, Newcastle Highly Specialised Mitochondrial Laboratory
16:45-17:00	GNAS methylation testing in a large cohort of patients with Pseudohypoparathyroidism (PHP) reveals overlap between the clinical subtypes	Simon Thomas, Wessex Regional Genetics Laboratory
17:00-17:15	Increasing diagnostic uplift in Familial Hypercholesterolemia through deep intronic LDLR interrogation	Celia Duff-Farrier, Bristol Genetics Laboratory
17:30-18:30	**? Drinks Reception @ the ICC**	

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Day 2 Programme

09:00-09:30	***Registration/Coffee/Trade/Posters***	
09:30	Introduction to meeting	
Session chair:		
09:35-09:55	Clinical Whole Genome Sequencing in Rare Disease: lessons learnt from the 100,000 Genomes Project and Genomic Medicine Service	Dalia Kasperaviciute, Head of Genomics Data Science, Genomics England
09:55-10:15	Diagnostic Discovery in the 100,000 Genomes Project	Susan Walker, Head of Translational Genomics, Genomics England
10:15-10:35	Application of ultra-rapid genotyping using LAMP coupled melt-curve analysis for DPYD testing	Laura Yarram-Smith, Bristol Genetics Laboratory
10:35-11:15	***Coffee/Trade/Posters* plus one 15 minute talk from Illumina**	
11:00-11:15	Improving paediatric and neonatal care for critically ill patients: A global overview of Illumina supported studies and collaborations	Maria Martinez-Fresno, Associate director of medical affairs GDT, EMEA, Illumina
Session chair:		
11:15-11: 30	RNAseq Panel Development for Haematology Oncology Gene Fusions and Partial Tandem Duplications	Sarah Hilton, Manchester Centre for Genomic Medicine
11:30-11:45	Molecular Testing of Solid Tumours: A Retrospective Review of Results Delivered by the CYSGODI Service (Cymru Service for Genomic Oncology Diagnoses)	Sian Wood, All Wales Medical Genomics Service
11:45-12:00	Evaluation of Illumina TSO500+HRD assay for the detection of Homologous Recombination Deficiency (HRD) in a cohort of Ovarian Cancer patients	Laura Yarram-Smith, Bristol Genetics Laboratory
12:00-12:15	Clinical Validity and Utility of ctDNA in the Management of High Grade Serous Ovarian Cancer	Djemilah Isakjee, West Midlands Regional Genetics Laboratory
	Lightning Presentation Session (8 total)	
12:25-12:30	Investigating the feasibility of the introduction of an ADTKD-MUC1 diagnostic service to the NHS Genomic Medicine Service	Emily Arbuthnot, Bristol Genetics Laboratory
12:30-12:35	Genomic testing in Phenylketonuria (PKU): National implementation of the Sapropterin NICE guidance	Rebecca Whittington, Bristol Genetics Laboratory
12:35-12:40	Cystic Renal Disease by WGS testing – the story so far in the GMS	Natalie Forester, Bristol Genetics Laboratory

12:40-12:45	Evaluation of the Clinical Utility of Optical Genomic Mapping in Diagnostic Haemato-Oncology Genetics	Laura Weir, Newcastle Genetics Laboratory
12:45-12:50	Potential for Long Read Sequencing Technology in Cancer Whole Genome Analysis	Helen Webb, Genomics England
12:50-12:55	A series of novel de novo microdeletions; expanding our knowledge of recently proposed AD neurodevelopmental disorder genes	Fiona Togneri, West Midlands Regional Genetics Laboratory
12:55-13:00	Solving the Unsolved: Reanalysis of genomic data to increase diagnostic yield in patients with critical illness and early-onset epilepsy	Jana Jezkova, All Wales Medical Genomics Service
13:00-14:00	***Lunch/Trade/Posters* plus two 15 minute talks from Qiagen and New England Biolabs**	
13:30-13:45	Overcoming the challenges of variant assessment and reporting	Dr Tim Bonnert, Director of Field Application Support, QIAGEN Digital Insights
13:45-14:00	DNA methylation analysis	***New England Biolabs presenting slot***
Session chair:		
14:00-14:30	2022 ACGS variant interpretation guidelines: aligning small variant and CNV classification	Emma Baple, Medical Director South West Genomic Laboratory Hub, Exeter Miranda Durkie, Lead Clinical Scientist, Sheffield Diagnostic Genetics Service Emma-Jane Cassidy, Principal Clinical Scientist, Wessex Regional Genetics Laboratory
14:30-14:45	Validation of whole genome sequencing analysis as a clinical test for cancer by comparison with standard of care test data	Susan Walker, Genomics England
14:45-15:00	UK somatic variant interpretation guidance (S-VIG)	George Burghel, Manchester Centre for Genomic Medicine
15:00-15:15	Reanalysis of exome sequencing data: The Exeter experience	Karen Stals, Exeter Genomics Laboratory
15:15-15:30	***Prizes, Closing Remarks and End of meeting***	