Association for Clinical Genomic Science

Summer Scientific Meeting 2023
11th – 12th September 2023, ICC, Birmingham

Day 1 Programme

09:00-10:00	***Registration/Coffee***	
09:50	Introduction to meeting	
10:00-10:50	The Newborn Genomes Programme – Navigating conditions and results including panel discussion	Amanda Pichini and Dr Harriet Etheridge, Genomics England
10:50-11:20	***Coffee/Trade/Posters***	
11:20-11:35	Development of a framework to support a culture of continuous quality improvement within the Exeter genomics laboratory	Melissa Sloman, Exeter
11:35-11:50	A strategic approach to NHS-Higher Education Partnerships, The Julia Garnham Centre	Duncan Baker & Adam Hodgson, Julia Garnham Centre, Sheffield
11:50-12:05	DECIPHER - Sharing phenotype-linked candidate diagnostic variants to enable rare disease diagnosis and research	Julia Foreman, DECIPHER
12:05-12:20	The likelihood ration calculator: bridging the Bayesian-Frequentist divide to enable flexibility in allocation of evidence weighting for PS4	Alice Garrett, Institute of Cancer Research
	Lightning Presentation Session	
12:30-12:35	Development and Validation of a Clinical Bioinformatics Pipeline for the Detection of CNVs in WGS	Joseph Halstead, Cardiff
12:35-12:40	CNV-classifier: A tool to improve CNV interpretation	Manuel Dominguez, Wessex
12:40-12:45	Whole genome sequencing increases diagnostic yield by improved CNV detection	Michael Spiller, Sheffield
12:45-12:50	Diagnostic confirmation of rare variant somatic mosaicism using droplet digital PCR	Joe Shaw, North Thames, London
12:50-12:55	Using protein meta-domains to increase data availability for missense variant classification	Adam Gunning, Exeter
12:55-13:00	Use of RNAseq to uplift the diagnostic yield in Primary Ciliary Dyskinesia	Simon Thomas, Wessex
13:00-13:05	Retrospective analysis of de novo phosphatase regulatory subunit gene variants in undiagnosed	Chris Armitage, Liverpool

	100kGP patients with rare developmental disorders	
13:05-13:10	The common PKD1 p.(Ile3167Phe) variant is hypomorphic and associated with severe, very early onset, biallelic polycystic kidney	Miranda Durkie, Sheffield
13:10-14:10	***Lunch/Trade/Posters***	
14:10-14:20	Sponsor talk 1	
14:20-14:30	Sponsor talk 2	
14:30-14:40	Sponsor talk 3	
14:40-14:50	Sponsor talk 4	
14:50-15:10	GENQA talk	TBC
15:10-15:45	ACGS AGM	
15:45-16:15	***Coffee/Trade/Posters***	
16:15-16:30	NHSE R14 Rapid Whole Genome Sequencing Service: Increasing diagnosis yield and reducing time to diagnosis for acutely unwell children	Karen Stals, Exeter
16:30-16:45	Diagnostic trio-based whole exome sequencing for severe developmental disorders in Scotland	Morad Ansari, Edinburgh
16:45-17:00	Somatic mosaicism identified during bespoke work-up for non-invasive prenatal diagnosis informs recurrence risk: proposal to develop an agreed panel of genes with known mosaicism risk to be funded through the National Genomic Test Directory for NIPD	Fiona McKay, North Thames, London
17:00-17:15	Assessment of bionano optical gene mapping - the Central and South Genomic lab hub experience	Anna Skowronska, Birmingham
17:15-17:30	Establishing a DNA methylation array service for rare disorders	Sarah Hilton, Manchester
17:30-18:30	**Drinks Reception @ the ICC**	

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Day 2 Programme

09:00-09:30	***Registration/Coffee/Trade/Posters***	
09:25	Introduction to meeting	
09:30-09:45	Non-coding variants in HK1 and hyperinsulinism: genotype-phenotype associations	Jayne Houghton, Exeter
09:45-10:00	Expanding the neurodevelopmental phenotype associated with HK1 de novo heterozygous missense variants	Rebecca Poole, Edinburgh
10:00-10:15	Biallelic HEATR5B variants cause pontocerebellar hypoplasia	Lewis Pang, Exeter
10:15-10:30	CYP2C19 genotyping to inform clopidogrel prescribing; results from a 1-year pilot	Matthew Beaney, Dundee
10:30-10:40	Sponsor Talk 5	
10:40-10:50	Sponsor Talk 6	
10:50-11:20	***Coffee/Trade/Posters***	
11:20-11:40	A de novo paradigm for sporadic genetic disease	Professor Joris Veltman, Dean of Biosciences Institute, Newcastle-upon-Tyne
11:40-12:10	GTAC presentation	Professor Kate Tatton-Brown, Clinical Director and Head of the Genomics Programme, Health Education England
	Lightning Presentation Session	
12:15-12:20	All Wales genomic sequencing service for intellectual disability and congenital malformations – A first year review	Sahra Warsome, Cardiff
12:20-12:25	The weird and wonderful: A 5 year evolution in the genetic testing for Beckwith Wiedemann and Russell-Silver syndromes	Melissa Connolly, Birmingham
12:25-12:30	WGS v small panel analysis for R58 Neurodegenerative disorders – has it improved diagnosis rate?	Claire Hodgkiss, Oxford
12:30-12:35	Identifying a suitable process for implementing and delivering a laboratory service for detecting mosaic TSC1 and TSC2 variants	Rosie Woodruff, Bristol

12:35-12:40	Learning from the diagnostic variants reported in the NHS GMS Whole Genome Sequencing Service for patients with Rare Disease	Dalia Kasperaviciute, Genomics England
12:40-12:45	The PHF21A neurodevelopmental disorder: an evaluation of clinical data from 13 patients	Rebecca Poole, Edinburgh
12:45-12:50	Utility of R14 testing in fetal samples with limited phenotypic information - diagnosis of TTC26-related biliary, renal, neurologic, and skeletal syndrome.	Ewa Goljan, Exeter
12:50-12:55	A five-year clinical audit for early gestation amniotic fluid samples	Manali Naik, Birmingham
12:55-13:00	Five-year review of the implementation of the molecular diagnostic pathway for Lynch Syndrome in people with colorectal cancer in NHS Tayside and NHS Highland health boards.	Christel Garcia-Petit, Dundee
13:00-14:00	***Lunch/Trade/Posters***	
14:00-14:15	The Development of an RNA Sequencing Protocol to Support the Transcriptomic Classification of Non-Muscle Invasive Bladder Cancer	Lauren Kettle, Birmingham
14:15-14:30	Whole genome sequencing to identify causative Fumarate Hydratase mutation in child with bilateral renal cell carcinoma	Chris Kershaw, Manchester
14:30-14:45	Low level BRCA2 pathogenic variant in lymphocyte DNA of a female patient seen at NWGLH	Simina Botosneanu, Manchester
14:45-15:05	Resolving the functional spectrum of pathogenic *POT1* alleles via saturation genome editing	Sofia Obolenski, Sanger Institute
15:05-15:25	Comprehensive and precise functional analysis of *RAD51C* by saturation genome editing at nucleotide resolution	Rebeca Olvera Leon, Sanger Institute
15:25-15:45	Timely cancer pathways	TBC
15:45-16:00	***Prizes, Closing Remarks and End of meeting***	

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Posters

Posters will be displayed in the foyer outside Hall 8. All authors must stand by their poster between 13:40-14:10 on Monday 11th September for questions.

Poster 1	Possible underdiagnosis of Temple Syndrome	Luke Redford, Wessex
Poster 2	Evaluating variant prioritisation tools for use in the Rapid Trio Fetal Exome Service at North Thames Genomic Laboratory Hub	Rachel Muir, North Thames, London
Poster 3	Genotype-phenotype correlation in Wilson Disease	Helen Patrick, Sheffield
Poster 4	"At home" patient-led capillary blood sampling allows equitable and timely access to genetic testing	Jayne Houghton, Devon
Poster 5	Audit of the NHS GMS Whole Genome Sequencing Service for patients with Rare Disease	Rachael Mein, NHS England
Poster 6	The utility of gene-agnostic trio analysis for the identification of novel biallelic forms of rare monoallelic disorders	Nicholas Head, Exeter
Poster 7	Review of the first 117 Solid Cancer Whole Genome Sequencing cases at WMRGL	Natasha Vafadar-Isfahani, Birmingham
Poster 8	The Julia Garnham Centre: Enhancing the student experience via pre-workforce engagement	Fleur Gardner, Julia Garnham Centre, Sheffield
Poster 9	Assay design and validation of a custom NGS panel as part of the TebeMRD clinical trial.	Kayleigh Mainwood, Oncology, Oxford
Poster 10	Analysis of factors determining success in FFPE based NGS panel testing for lung and ovarian cancer in the NHS	Priyanka Bhattacharya, Manchester
Poster 11	Mainstreaming saliva testing for Familial Hypercholesterolaemia diagnostics	Kate Annesley, Cambridge
Poster 12	RNA PanCancer fusion detection with Arriba/Starfusion. An effective and affordable solution.	Martin van der Linde,