Association for Clinical Genomic Science

Annual Scientific Meeting 2021
29th – 30th September 2021

Day 1 Programme

09:20	**Introduction to meeting**	
09:30-09:50	Nineteen million samples tested for SARS-CoV-2 RNA in 18 months: The Lighthouse Laboratory, UK Biocentre, Milton Keynes experience on mass testing	Dr Malur Sudhanva, Clinical Advisor, Milton Keynes Covid Testing Lighthouse Laboratory
09:50-10:10	Utilising genomic data within the National Disease Registration Service to improve understanding and patient outcomes in cancer, congenital anomalies and rare disease	Dr Steven Hardy, Head of Molecular Diagnostics, National Disease Registration Service, Public Health England
10:10-10:30	The UK Biobank as a resource to investigate reproductive success in 500,000 humans	Eugene Gardner, Wellcome Sanger Institute, Cambridge & MRC Epidemiology Unit, University of Cambridge
10:30-11:15	**Break: Sponsor talks**	
10:30-10:40	The Next Generation of Targeted Methylation Sequencing	Bryan Hoglund, Senior Business Development Manager, NGS at TWIST Bioscience
10:40-11:00	Consistent interpretation of CNVs to increase diagnostic yields	Ruth Burton, Senior Scientist – Informatics, Qiagen
11:05-11:15	**Feedback from the ACGS Scientific and Technology committee**	
11:15-11:30	Development of a diagnostic service for extended characterisation of apparently de novo disease associated variants	Jonathan Williams, Oxford
11:30:11:45	Optimisation and validation of droplet digital PCR (ddPCR) as a technique for detecting mitochondrial depletion and deletions	Ruth Braham, GOS, London
11:45-12:00	Development of a next generation sequencing assay for RPGR ORF15	Caitlin Fleming-Knox, Manchester
12:00-12:15	Genomic testing in patients with suspected familial chylomicronaemia syndrome (FCS) and evaluation of the FCS clinical scoring tool	Sarah Marsh, Bristol
12:15-12:30	Preliminary study validating the 12-SNP LDL-C polygenic risk score for familial hypercholesterolaemia in London lipid clinics	Krunal Mahida, GOS, London
12:30-12:45	Implementation and audit of the DPYD service in the West Midlands Regional Genetics Laboratory	Lowri Hughes, Birmingham
12:50-13:50	**Lunch: Sponsor talks**	
12:50:13:10	MSI analysis by NGS using a small panel of markers without paired normal tissue	Dr Richard Talbot, Field Application Support for Celemics Inc (VH Bio)

13:10-13:30	The voices of patients in the diagnostic journey	Shirlene Badger, Patient Advocacy Lead, EMEA, Illumina
13:30-13:50	Putting the patient at the heart of patient care	Dr Charles Steward, Patient Advocacy and Engagement Lead, Congenica
13:55-14:05	**Feedback from the ACGS Quality committee**	
14:05-14:35	Guidelines for the classification of variants in non-coding regions	Dr Jamie Ellingford, University of Manchester and Nicky Whiffin, Wellcome Centre for Human Genetics, University of Oxford
14:35-14:45	MRSD: a novel quantitative approach for assessing suitability of RNA-seq in the investigation of mis-splicing in Mendelian disease	Dr Jamie Ellingford, University of Manchester
14:45-15:15	Familial hypercholesterolaemia – Evolving genomic practice to meet the NHS Long Term Plan	Maggie Williams, Bristol
15:15-15:40	**Break: Sponsor talk**	
15:15-15:35	Clinical utility of microsatellite instability (MSI) testing for Lynch syndrome and anti-PD1 immunotherapy for colorectal cancer	Victoria Wilson, Global Clinical Collaborations manager, Promega
15:40-16:20	Variable variants – how do we standardise?	Becky Treacy, Jenni Fairley and Melody Tabiner - GENQA
16:20-16:35	Successful repatriation of genetic testing services to the North East of Scotland	Dawn O'Sullivan, Aberdeen
16:35-16:50	Implementing the national rare disease WGS service: East GLH perspective	Rachel Challis, Cambridge
	Close of Day one	

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Day 2 Programme

09:20	***Introduction to Meeting***	
09:30-09:50	Clinical Applications of nanopore sequencing	Chris Watson, Leeds
09:50-10:10	SE-HMDS and SESGS Bionano Experience	Dr Azim Mohamedali, Kings College, London & Hannah Heppell, Edinburgh
10:10-10:30	Long Read Sequencing for Novel Diagnostic Tests in Cancer and Rare Disease	Dr Ali Awan, Guy's & St Thomas'
10:30-11:15	**Break: Sponsor talks**	
10:30-10:50	NEXTFLEX® Solutions: From targeted gene panels to whole genome sequencing	Raul Gonzalez, Sales Specialist - Applied Genomics, Perkin Elmer
10:50-11:10	UMI-enabled sequencing of full length B-cell and T-cell receptor repertoires for understanding immune response to human disease	Adam Peltan, Senior Technical Applications Specialist - NGS and Customised Solutions, NEB
11:15-11:25	**Feedback from the ACGS Workforce and Development committee**	
11:25-11:40	Introduction of the renal cell carcinoma microarray service in the North of Scotland Genetics and Molecular pathology laboratory	Myra Loomer, Aberdeen
11:40-11:55	Limit of detection assessment of the ArcherDx RNA Pan Cancer panel	Charlotte Flanagan, Guy's, London
11:55-12:10	Detection of clinically relevant genetic variants in neuroblastoma	Jack Baines, Newcastle
12:10-12:25	Digital pathology and automated macrodisssection	Jason Parr, Manchester
12:25-13:30	**Lunch: Sponsor talks**	
12:25-12:45	Can a Genomic LIMS improve efficiency in the laboratory?	Emma Huntridge, Genetics Business Development Director, CliniSys
12:45-13:05	Improved human exome sequencing workflow with the most complete coverage	Peter Verhasselt, Senior Field Applications Scientist NGS, IDT
13:30-14:15	***Lightning Presentation Session (8 total)***	
	Reanalysis for the 100,000 Genomes Project Participants: Completing the clinical picture	Sophia Donavan, Liverpool
	Scottish Genomes Partnership (SGP): The North East of Scotland perspective	Christine Armstrong, Aberdeen
	Investigating the potential of AWS cloud computing services for clinical bioinformatic pipelines	Rebecca Sadler, Oxford

	Implementation of a national pharmacogenetics testing service for DPD variants in Wales – first year review	Elin Brier, Cardiff
	Implementation of a NGS based fusion testing service for solid tumours at the Aberdeen Medical Genetics Laboratory	Katarzyna Matula, Aberdeen
	Benefits and implications of Next Generation sequencing in Haemato-Oncology	Paula Scott, Aberdeen
	All Wales Medical Genomics Service – Transforming Welsh Cancer Genomics – Implementation of a comprehensive DNA and RNA next generation sequencing panel for solid and haematological cancers	Megan Fealey, Cardiff
14:15-14:45	Newborn WGS	Dr Richard Scott, Clinical Director, Genomics England
14:45-15:10	**Break: Sponsor talk**	
14:45-15:05	Advancing on a Sure Path: Eliminating sample prep bottlenecks in Clinical Genetic Labs	Sammi Alouni, Service Manager & Principal Clinical Scientist for the Liver Molecular Genetics Laboratory, Viapath, Kings College Hospital, and Dr Michelle Hiscutt, Product Specialist, Agilent
15:10-15:20	**Feedback from the ACGS Communications committee**	
15:20-15:50	Genetics of Obesity: Can an old dog teach us new tricks?	Dr Giles Yeo, University of Cambridge
15:50-16:20	Mitochondrial genomics in clinical practice	Professor Robert Taylor, Newcastle
16:20-16:45	**ACGS AGM & prizes**	
	Closing Remarks and End of meeting	