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	Genomic test data collected at the National Disease Registration Service	Steven Hardy, Public Health England
09:50-10:10	Covid testing	TBC
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	Experience with the Bionano platform	Dr Azim Mohamedali, Kings College, London
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

	Identification of a rare chromosomal insert as the cause of X-linked hypophosphatemic rickets	Sean Hegarty, Belfast
	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	