Association for Clinical Genetic Science
Part of the British Society for Genetic Medicine

Summer Scientific Meeting
Rare & Reproductive Genomics & Applying Genomics to Cancer
4th – 5th of July 2016, Austin Court, Birmingham

Day 1 Programme: Rare & Reproductive Genomics

09:00 – 09:30	***Registration/Coffee***	
09:30 – 10:15	**ACGS AGM - Welcome and Chair: Dom McMullan**	
10:15 – 10:45	***Coffee***	
10:45 – 11:45	**Data Sharing/Collation initiatives Chair: Dom McMullan**	
	Incorporation of Genetic Data into PHE'S National Disease Registration Databases	Fiona McRonald, PHE
	DECIPHER: new tools and plans for transition to new genome assembly (build 38)	Matthew Hurles, Sanger Institute
	The regulatory landscape and its impact on practice	Alison Hall, PHG Foundation
11:45 – 13:00	**100, 000 Genomes - Chair: Val Davison**	
	Validation of 100,000 Genomes results; from WGS to the Genomics England result	Emma Baple, Genomics England
	UKGTN/ PanelApp Utility	Fiona Macdonald, UKGTN
	Phenotype-aware approaches to variant prioritisation	Damian Smedley, Genomics England
	Validation of 100,000 Genomes results; from Genomics England result to the clinical diagnostic report	Sian Ellard, Exeter
13:00 – 13:50	***Lunch***	
	Early afternoon session – Chair: Maggie Williams	
13:50 – 14:35	**Key note speaker: Why there is more gene evolution than protein function**	Lawrence Hurst, University of Bath
14:35 – 14:50	Role of the clinical review panel in the PAGE study	Eamonn Maher, Cambridge
14:50 – 15:05	Setting standards and Raising Quality for Clinical Bioinformatics	Ahn JooWook, Guy's
15:05 – 15:45	**Challenges in Variant Interpretation (1) Chair: Sian Morgan**	
	What lies beneath: challenges in reporting SNP microarray results	Jonathan Waters, GOS
	Results of NIPT confirmation testing, following invasive prenatal sampling, highlight a need for consistent NIPT provision in the NHS	Fiona Togneri, Birmingham
	Copy number variants of uncertain significance in prenatal diagnosis, are the goal posts moving?	Lisa Burvill-Homes, Bristol
15:45 – 16:15	*Coffee*	
16.15 – 17:45	**Challenges in variant interpretation (2) Chair: Simon Thomas**	
	Putative low penetrance or susceptibility variants: sodium channel genes in painful neuropathy as an example	Carl Fratter, Oxford
	Challenges and solutions to variant detection and interpretation in clinical exome sequencing; lessons from the first 150 cases reported	Thomas Cullup, GOS

	Making non(sense) of variant interpretation for patients tested on a 30 gene Next Generation Sequencing (NGS) panel for Disorders of Sex Development (DSD)	Nick Drinkall, Birmingham
	Cryptic splice sites and micro-exons – A case study: MYBPC3 c.1224-52G>A in association with hypertrophic cardiomyopathy	Helen Sage, Oxford
	To cascade or not to cascade? Novel approaches to VUS classification in FH	Rebecca Whittington, Bristol
	Rising to the challenges of variant interpretation for the diagnosis of rare disease by exome sequencing	Sian Ellard, Exeter
	Reclassification of variants: Implications from the cardiac perspective	Karen McGuire, Oxford
	Genomic Evaluation Meetings – effective reporting of challenging variants	Olivera Spasic-Boskovic, Cambridge
17:45 – 18:00	**Closing Remarks: Dom McMullan**	
18:00 – 19:30	**Drinks Mixer**	

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