Summer Scientific Meeting 2017
26th – 27th of June 2017, Austin Court, Birmingham

Day 1 Programme: WGS Implementation & Professional initiatives

09:00 – 09:30	*Registration/Coffee/Posters*	
09:30 – 10:00	Creating the NHS Genomic Medicine Service	Professor Sue Hill OBE (NHSE)
10:00 – 10:20	100,000 genomes - Rare Disease Programme update	Richard Scott (Genomics England)
10:20 – 10:40	100,000 genomes - Cancer Programme update	Clare Turnbull (Genomics England)
10:40 – 11:00	NHS Fetal Anomaly Screening Programme	Annett McHugh (PHE)
11:00 – 11:20	Q + A	Above Speakers
11:20 – 11:50	*Coffee/Trade/Posters*	
11:50 – 12:05	The Academy for Health Care Science (AHCS)	Professor Brendan Cooper (AHCS)
12:05 – 12:25	Health Education England - Genomics Education Programme	Anneke Seller (HEE)
12:25 – 13:00	CEQAS and NEQAS overview and update	Ros Hastings / Katrina Rack (CEQAS), Jenni Farley /Farrah Khawaja (NEQAS)
13:00 – 14:00	*Lunch/Trade/Posters*	
14:00 - 14:40	Improving the interpretation of rare variants through inter-disciplinary working	Professor Sian Ellard (Royal Devon & Exeter) / Thalia Antoniadi (Birmingham Women's & Children's) / Emma Baple (Royal Devon and Exeter)
14:40 – 15:00	Validation and Reporting Working Groups (RD and Cancer)	Dom McMullan (Birmingham Women's & Children's) / Chris Wragg (North Bristol)
15:00 – 15:15	Evaluation of a modified Genomics England (GEL) Tiering System for NGS Variant Analysis	Simon Thomas (Salisbury)
15:15 – 15:30	ACGS Bioinformatics Group Update	Joo Wook Ahn (Guy's & St Thomas')
15:30 – 16:00	*Coffee/Trade/Posters*	
16:00 – 16:15	'Dynamic Panelisation' as an approach to clinical exome analysis	David Gokhale (Central Manchester University Hospital)
16:15 – 16:30	A targeted exome testing strategy for inherited Intellectual Disability	Stephanie Barton (Central Manchester University Hospital)
16:30 – 17:00	The Genetics Clinic of the Future – a Dutch consortium approach to diagnostic Whole Genome Sequencing	Ies Nijman (UMC Utrect)
17:15 – 18:00	ACGS AGM	
18:00 – 19:30	*Drinks Reception*	

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Day 2 Programme: Challenges in variant interpretation

09:00 – 09:30	*Registration/Coffee/Poster*	
09:30 – 09:50	Whole genome and transcriptome analysis in anaplastic meningioma	Patrick Tarpey (Addenbrookes)
09:50 – 10:10	Genomic Profiling in Follicular Lymphoma and Implications for Precision Medicine	Shamzah Araf (Barts Cancer Institute)
10:10 – 10:25	RNA-Sequencing to detect gene fusions in acute leukaemia: A proof-of-principle study	Melissa Connolly (Birmingham Women's & Children's)
10:25 – 10:40	Nanopore long read sequencing for detection of point mutations and structural variants	Kezia Brown (Viapath)
10:40 – 10:55	Rapid clinical NGS response to containing infectious disease outbreaks	Kim Brugger (Addenbrookes)
11:00 – 11:30	Coffee/Trade/Posters	
11:30 – 11:45	Non-invasive prenatal testing (NIPT) for fetal aneuploidy – Challenging cases and reporting conundrums!	Kirsten McKay Bounford (Birmingham Women's & Children's)
11:45 – 12:00	Challenges in interpretation of relative haplotype dosage analysis (RHDO) data; our experience in implementing a non-invasive prenatal diagnosis (NIPD) clinical service	Beth Young (Birmingham Women's & Children's)
12:00 – 12:15	Automation of bioinformatics analysis to improve efficiency of Non-Invasive Prenatal Diagnosis for monogenic disorders in North East Thames Regional Genetics	Helena Ahlfors (Great Ormond Street)
12:15 – 12:30	Reproductive options for patients with mitochondrial DNA disease: Using mitochondrial donation to prevent disease transmission	Emma Watson (Newcastle)
12:30 – 12:45	Challenges and complexities in the interpretation of mitochondrial DNA (mtDNA) sequence variants	Steven Hardy (Newcastle)
13:00 – 14:00	Lunch/Trade/Posters	
14:00 – 14:40	Causes and consequences of de novo mutations in health and disease	Professor Joris Veltman (Newcastle University)
14:40 – 15:00	Establishing variant pathogenicity in inherited cardiac conditions	Nicky Whiffin (Imperial College London)
15:00 – 15:15	A comprehensive next generation sequencing service for inherited cardiac conditions; a review of the first three years of implementation	James Eden (Central Manchester University Hospital)
15:15 – 15:30	Exome sequencing identifies a new form of autosomal recessive Spondylocostal/spondylothoracic dysostoses	Melissa Sloman (Royal Devon & Exeter)

15:30 – 15:45	Rapid analysis of whole-genome sequence of children presenting to neonatal and paediatric intensive care	Alba Sanchis (University of Cambridge)
15:45 – 16:15	*Coffee/Trade/Posters*	
16:15 – 16:35	Using large-scale human genetic variation to inform variant prioritization in neuropsychiatric disorders	Kaitlin Samocha (Wellcome Trust Sanger Institute)
16:35 – 16:50	Comprehensive rare variant analysis via whole-genome sequencing to determine the molecular pathology of inherited retinal disease	Keren Carss (University of Cambridge)
16:50 – 17:05	Updating penetrance estimates for syndromes with variable phenotypic manifestation	Adele Corrigan (Viapath)
17:05 – 17:20	Lessons from DDD study results: when “de novo, loss of function” doesn’t equal pathogenic	Sarah Turton (Birmingham Women's & Children's)
17:20 – 17:35	The clinical utility of functional mRNA evaluation of rare genetic variants in diagnostic practice	Celia Duff-Farrier (North Bristol)
	Closing remarks	

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