Summer Scientific Meeting 2019
10th – 11th June 2019, ICC, Birmingham

Day 1 Programme

09:00-09:30	***Registration/Coffee/Posters***	
09:30	Introduction to meeting	
09:40-10:00	Phenotypic Spectrum of neurodevelopmental CNVs	Dr Kimberley Kendall (MRC Centre for Neuropsychiatric Genetics and Genomics, Cardiff)
10:00-10:30	Clinical Implementation of Whole Genome Sequencing	Dr Madhuri Hegde (Adjunct Professor of Human Genetics and Pediatrics, Emory University and Georgia Institute of Technology; VP & CSO PerkinElmer Global Laboratories)
10:30-11:00	***Coffee/Trade/Posters***	
11:00-11:20	CNS tumours, the new WHO and the impact of molecular genetics	Professor Federico Roncaroli (University of Manchester)
11:20-11:40	ESMO germline focused analysis of tumour-only sequencing	Dr Clare Turnbull (Institute of Cancer Research)
11:40-12:00	Data interpretation in cancer genomics	Kevin Litchfield (The Francis Crick institute)
12:00-12:20	Update on somatic variants	Christopher Wragg (Bristol)
12:20-13:00	***Lightning Presentation Session (8 total)***	
	Methylation Sensitive High Resolution Melting (MS-HRM) Assay for the Detection of BRCA1 and BRCA2 Promoter Hypermethylation	Gareth Gerard (London)
	The detection of novel Alu insertions in cancer susceptibility genes	Evgenia Petrides (Oxford)
	Preparing Solid Tumour Reports for the Genomic Era	Sian Lewis (Wales)
	Somatic Variant Interpretation	Sian Lewis (Wales)
	Longitudinal changes in mutational burden associated with disease progression in myeloid malignancies	Jan Taylor (Leeds)
	Review of FFPE tissue testing in patient with Colorectal Cancer	Helene Schlecht (Manchester)
	Histopathological assessment training for Genomics Clinical scientists	George Burghel (Manchester)
	Assimilation of high-resolution HLA alleles from low-resolution serological typing; a computational approach.	Adriana Toutoudaki (Cambridge)
13:00-14:00	***Lunch/Trade/Posters***	

14:00-15:00	ACGS AGM	
15:00-15:20	GENQA – variant interpretation	Dr Sandi Deans
15:20-15:40	EMQN – NGS sequencing scheme	Dr Simon Patton
15:40-16:10	***Coffee/Trade/Posters***	
16:10-16:25	WMRGL integrated reporting of targeted NGS and cytogenetic results in AML and MDS: review of the first 1,000 cases.	David Hill (Birmingham)
16:25-16:40	Identifying potential germline variants during somatic testing: a selection of challenging cases from the WMRGL Haemato-Oncology	Kimberley Reay (Birmingham)
16:40-16:55	100,000 Genomes Project: Validating Whole Genome Sequencing (WGS) for clinical use in Cancer Programme	Alona Sosinsky (Genomics England)
16:55-17:10	The analysis of clinically relevant structural variants in 100K cancer genomes	Jamie Trotman (Cambridge)
17:10-17:25	Increased diagnostic yield from gene-agnostic trio analysis of 100,000 Genomes Project sequence data	Karen Stals (Exeter)
17:30-19:00	**Drinks Reception**	

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Day 2 Programme

09:00-09:30	***Registration/Coffee/Trade/Posters***	
09:30-09:50	Uncovering pathogenic variants in the non-coding genome	Dr Jamie Ellingford (University of Manchester)
09:50-10:10	5' untranslated regions, upstream open reading frames and translational regulation	Dr Nicola Whiffin (Imperial College London)
10:10-10:25	Pathogenic non-coding variants in inherited retinal disease from the 100,000 genome project	Dr Kate Oprych (Great Ormond Street Children's Hospital)
10:30-11:00	***Coffee/Trade/Posters***	
11:00-11:15	NIPT as part of the national screening programme in Wales – a 1 year review	Sarah Anderson (Wales)
11:15-11:30	Non-invasive diagnosis of retinoblastoma using cell-free DNA from aqueous humour	Amy Gerrish (Birmingham)
11:30-11:45	Non-Invasive Prenatal Diagnosis for Sickle Cell Disease by Droplet Digital PCR and Next Generation Sequencing	Joe Shaw (GOS, London)
11:45-12:00	Exome sequencing of 875 parental/fetal trios with structural abnormalities revealed by ultrasound in the UK prenatal assessment of genomes and exomes (PAGE) project	Dominic McMullan (Birmingham)
12:00-12:15	A pilot study for the application of Next Generation Sequencing in Cystic Fibrosis Newborn Screening	Richard Kirk (Sheffield)
12:15-12:30	Genetic testing and variant classification in Primary Ciliary Dyskinesia reveals considerable genetic heterogeneity despite a high proportion of homozygous cases	Simon Thomas (Salisbury)
12:30-13:10	***Lightning Presentation Session (8 total)***	
	Cost effective screening of the PKD1 and PKD2 genes in patients with autosomal dominant polycystic kidney disease (ADPKD)	Lewis Darnell (Nottingham)
	Focused Exome Pool-Seq of parent offspring Trios in previously negative cases	Kathleen Murphy (Manchester)
	Assessing whole genome sequencing as a diagnostic test for mitochondrial disease.	Frankie Macrae (Cambridge)
	Whole genome sequencing in a series of Primary Ciliary Dyskinesia cases identifies molecular diagnoses in DNAH5	Holly Black (Edinburgh)
	The 100,000 Genomes Project; successful cases	Amy Webb (Salisbury)

	and cautionary tales – the Wessex experience	
	Targeted microarray design increases detection of clinically-relevant variants across multiple NHS genomics centres	Jana Jezkova (Wales)
	Detection of Microdeletion Syndromes by NIPT	Kathryn Jones (Liverpool)
	A new diagnostic approach to calling CNVs from low read-depth genome sequencing data	Bethany Wild (Guy's, London)
13:10-14:40	***Lunch/Trade/Posters*** **Poster viewing session 13:30-14:00**	
14:40-15:10	Genetic Risk Prediction	Professor Andrew Read (University of Manchester)
15:10-15:40	Genomic testing in a population setting in biobank samples	Dr Caroline Wright (University of Exeter)
15:40-15:55	A process for gaining consensus gene panels in the Genomic Medicine Service using PanelApp	Ivone Leong (Genomics England)
15:55-16:10	CNV and SNV analysis of 844 intellectual disability genes in singleton referrals	Hannah Grayton (Cambridge)
16:10-16:25	DECIPHER - Enabling the sharing and interpretation of rare disease variants and associated clinical phenotypes	Julia Foreman (Welcome Sanger Institute)
	Closing Remarks and End of meeting	

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