The Association of Clinical Pathologists

acgs 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 2 Programme: Applying Genomics to Cancer

09:00 – 09:30	***Registration/Coffee***	
	Welcome: David Baty	
09:30 – 10:40	**100, 000 Genomes – Chair: Shirley Henderson**	
	Validation in 100,000 genomes cancer programme	Shirley Henderson, Oxford
	Cancer sample quality	Louise Jones, Barts, London
	100,000 genomes cancer programme: A GMC perspective	Emily Shaw, Wessex Genomic Medicine Centre
	100,000 Genomes Cancer Programme Discussion Session	
10:40 – 11:40	**Molecular Pathology – Chair: David Baty**	
	CRUK questionnaire of solid tumour services	Sumi Subramaniam, Cancer Research UK
	Diagnostic ctDNA analysis in NSCLC	Angharad Williams, Cardiff
	NGS to detect CNVs and gene fusions in CR-UK SMP2	Michelle Wood, Cardiff
11:40 – 12:10	**Coffee**	
12:10 – 13:10	**Challenges in variant interpretation – Chair: Christine Waterman**	
	CR-UK Stratified Medicine Programme: Challenge of interpretation and reporting in solid tumours	Pauline Rehal, Birmingham
	Separating the Wheat from the Chaff; The Challenges of Interpreting Multiple Variants in Familial Cancer Patients	Tina Bedenham, Oxford
	A complex NF2 mutation detected by custom Next Generation Sequencing (NGS) analysis	Michael Bulman, Manchester
	Characterising retrotransposon insertion mutations in Neurofibromatosis type 1 (NF1)	Naomi Bowers, Manchester
	Characterisation of co-operating genetic lesions to aid the clinical management of AML patients	Vera Martin Dias, Birmingham
13:10 – 14:10	**Lunch**	
14:10 – 15:30	**UK NEQAS – Chair: Sandi Deans**	UK NEQAS Molecular Genetics
	How close are we to standardised extended RAS gene mutation testing? The UK NEQAS evaluation	Susan Richman, Leeds
	Introducing an EQA for testing ctDNA in plasma	Sandi Deans, Edinburgh
	Measuring DNA – not so simple	Nelson Iley, Edinburgh
	How low can you go? A snapshot of molecular	Jenni Fairley, Edinburgh

	pathology test sensitivity and specificity	
15:30 – 16:00	**Coffee**	
16:00 – 17:15	**NGS testing in Haemato-oncology – where are we going? Chair: Alastair Reid**	
	Integration of multi-gene analysis for myelodysplastic syndromes (MDS) and acute myeloid leukaemia (AML) using the Illumina Trusight Myeloid panel	Kate Pearce, Newcastle
	Development of a Next Generation Sequencing (NGS) multi-locus immunoglobulin (IgH) and T-cell receptor (TCR) approach for use in Minimal Residual Disease (MRD) detection in Childhood Acute Lymphoblastic Leukaemia (ALL)	Stephanie Wakeman, Bristol
	The discovery of low level TP53 variants using a custom designed NGS panel and the impact on clinical outcome	Ria Hipkiss, Oxford
	What is the Clinical Utility of a Next Generation Sequencing (NGS) Myeloid Panel Test?	Adele Timbs, Oxford
	Validation of a capture-based NGS panel for mutation profiling (including FLT3-itd and KMT2A gene fusions) in AML patients	Paula Page, Birmingham
	Implementation of NGS into Diagnostic Practice for Myeloid Malignancies: Experience from HMDS	Jan Taylor, Leeds
17:15 – 17:30	**Closing Remarks**	

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