

British Society for Genetic Medicine Annual Conference - UK Clinical Genomics 2019 -

Thursday 10th October 2019 | Royal College of Physicians | Regent's Park | London | NW1 4LE

09:00 – 09:55	Registration <i>Wolfson Theatre foyer</i>	
	Refreshments and sponsor exhibition <i>Osler and Long Rooms, First floor</i>	
09:55 – 10:00	Welcome <i>Wolfson Theatre</i>	Anneke Lucassen (<i>Chair, BSGM</i>)
10:00 – 12:10	UK Clinical Genomics <i>Chairs: John Burn and Emma Woodward</i> <i>Wolfson Theatre</i>	
10:00 – 10:30	Implementing pharmacogenomics into clinical practice	Munir Pirmohamed (<i>The University of Liverpool</i>)
10:30 – 10:50	Transforming Genomic Medicine Initiative (TGMi) session: Incorporating protein structure into variant interpretation	Caroline Wright (<i>University of Exeter</i>)
10:50 – 11:10	Working for speedy, accurate, stable, and clear genetic test reports	David Fitzpatrick (<i>The University of Edinburgh</i>)
11:10 – 11:30	Structural variants and their effect on gene regulation	Stefan Mundlos (<i>Max Planck Institute for Molecular Genetics, Berlin</i>)
11:30 – 11:50	Interpreting beyond the exome: variants in 5' untranslated regions result in loss of protein translation and cause human disease	Nicky Whiffin (<i>Imperial College London</i>)
11:50 – 12:10	The splice of life – insights into genetic disease	Diana Baralle (<i>University of Southampton</i>)
	Lunch and Trade Exhibition <i>Osler and Long Rooms, First floor</i>	
	Lunchtime meetings	
12:10 - 12:15	The new curriculum for SpR training in Clinical Genetics	Wolfson Lecture Theatre
12:15 – 12:45	UK Fetal Genomics Steering Group Meeting	Wolfson Lecture Theatre
12:30 – 13:00	NIHR Early Career Researchers Meeting: An idiot's guide to getting started on 100,000 genomes analysis (Dr Tim Hearn, Prof Eamonn Maher)	Dorchester Library
13:10 – 14:45	Concurrent submitted abstract sessions <i>Wolfson Theatre</i>	Dorchester Library (first floor)
13.10	Onasemnogene Aβparvovec Gene-Replacement Therapy for Spinal Muscular Atrophy: From Bench to Bedside. Imran Kausar.	Blood methylation signatures specific for individual developmental disorders enable resolution of 90% of variants of uncertain significance in these genes. Juliet E. Handsaker.
13.25	Exome sequencing reveals de novo mutations and deletions in severe unexplained male infertility. Joris Veltman.	A restricted spectrum of KMT2D variants cause a multiple malformations disorder distinct from Kabuki syndrome. Verity Hartill.
13.40	Familial Hypercholesterolaemia: Shared experience from 10,000 cases to enhance testing in the genomic medicine service. Elizabeth Watson.	Incomplete annotation has a disproportionate impact on our understanding of Mendelian and complex neurogenetic disorders. David Zhang.
13.55	Non Invasive Prenatal Testing (NIPT): What do the results mean for our patients and what testing should we be recommending? Fiona Togneri.	De novo variants in SLC12A2 cause a neurodevelopmental disorder by reducing NKCC1 function. Alisdair McNeill.
14.10- 14:45 (Lightening presentations to run concurrently)	Hereditary Leiomyomatosis and Renal Cell Cancer (HLRCC): Clinical, molecular and surveillance features in a cohort of 185 affected individuals. Claire Forde.	Prenatal exome sequencing in unexplained fetal oedema: a game changer. Esther Dempsey
	Structural Aberrations with Secondary Implications (SASIs): consensus recommendations for reporting of cancer susceptibility genes identified during analysis of Copy Number Variants (CNVs). Sabrina Talukdar.	Early onset polycystic kidney disease associated with functionally significant PKD1 hypomorphic variants. Miranda Durkie.
	Molecular diagnosis of mosaic disease in neurofibromatosis type I. Emma Burkitt Wright.	De novo missense variants in WDR37 associated with syndromic coloboma; findings from the 100,000 genomes project. Eleanor Hay.

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BRCA1/2 genetic testing in a series of women with Uterine Serous Cancer. **Rachel Jones.**

Null variants and deletions in BRWD3 cause an X-linked syndrome of mild-moderate intellectual disability, macrocephaly, and obesity: a series of 17 patients. **Philip Ostrowski.**

Direct-to-consumer genetic test results: cautionary tales. **Rachel Horton.**

Analysis of 1514 Rare Disease 100,000 Genomes Project cases in the South West of England Genomic Laboratory Hub (SWGLH) – learning from positive diagnoses to enhance the Genomic Medicine Service. **Hannah Robinson.**

Parent and health professional experiences and views of genome sequencing for rapid diagnosis in critically ill children. **Melissa Hill.**

Clinical and genetic variability in children with partial albinism. **Patrick Campbell.**

14:45 – 15:10

Afternoon Tea and Trade Exhibition
Osler and Long Rooms, First floor

15:10 – 17:30

Applications of Genomics in Management of Human Disease
Chairs: Eamonn Sheridan and Carol Gardiner
Wolfson Theatre

15:10 – 15:40

Finding the origins of childhood cancer.

Sam Behjati (*Sanger Centre*)

15:40 – 16:10

Experience as knowledge: reproductive decisions of families living with genetic disease.

Felicity Boardman (*University of Warwick*)

16:10 – 16:40

Lessons and opportunities from curating the genotypes of half-a-million people in the UK Biobank.

Clare Bycroft (*UK Biobank*)

16:40 – 17:30

The BSGM Lecture 2019

From syndromology to population cancer risk stratification: a life's journey in cancer genetics

Gareth Evans (*The University of Manchester*)

17:30 – 18:00

BSGM Annual General Meeting