

# British Society for Genetic Medicine Annual Conference

## - UK Clinical Genomics 2019 -

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

09:00 – 09:55	<b>Registration and Coffee</b>	
09:55 – 10:00	<b>Welcome</b>	<b>Anneke Lucassen</b> ( <i>Chair, BSGM</i> )
10:00 – 12:10	<b>UK Clinical Genomics (Wolfson Theatre)</b>	
10:00	Implementing pharmacogenomics into clinical practice	<b>Munir Pirmohamed</b> ( <i>The University of Liverpool</i> )
10:40	<b>Transforming Genomic Medicine Initiative (TGMI) session:</b> Incorporating protein structure into variant interpretation	<b>Caroline Wright</b> ( <i>University of Exeter</i> )
11:00	Working for speedy, accurate, stable, and clear genetic test reports	<b>David Fitzpatrick</b> ( <i>The University of Edinburgh</i> )
11:20	Structural variants and their effect on gene regulation	<b>Stefan Mundlos</b> ( <i>Max Planck Institute for Molecular Genetics, Berlin</i> )
11:30	Interpreting beyond the exome: variants in 5' untranslated regions result in loss of protein translation and cause human disease	<b>Nicky Whiffin</b> ( <i>Imperial College London</i> )
11:50	The splice of life –insights into genetic disease	<b>Diana Baralle</b> ( <i>University of Southampton</i> )
12:10 – 13:10	<b>Lunch and Trade Exhibition</b>	
13:10 – 14:45	<b>Concurrent submitted abstract sessions</b> <b>Wolfson Theatre</b>	<b>Dorchester Library</b>
13:10	Onasemnogene Aboaparvovec Gene-Replacement Therapy for Spinal Muscular Atrophy: From Bench to Bedside. <b>Imran Kausar.</b>	Blood methylation signatures specific for individual developmental disorders enable resolution of 90% of variants of uncertain significance in these genes. <b>Juliet Handsaker.</b>
13:25	Exome sequencing reveals de novo mutations and deletions in severe unexplained male infertility. <b>Joris Veltman.</b>	A restricted spectrum of KMT2D variants cause a multiple malformations disorder distinct from Kabuki syndrome. <b>Verity Hartill.</b>
13:40	Familial Hypercholesterolaemia: Shared experience from 10,000 cases to enhance testing in the genomic medicine service. <b>Elizabeth Watson.</b>	Incomplete annotation has a disproportionate impact on our understanding of Mendelian and complex neurogenetic disorders. <b>David Zhang.</b>
13:55	Non Invasive Prenatal Testing (NIPT): What do the results mean for our patients and what testing should we be recommending? <b>Fiona Togneri.</b>	De novo variants in SLC12A2 cause a neurodevelopmental disorder by reducing NKCC1 function. <b>Alisdair McNeill.</b>
14:10	Hereditary Leiomyomatosis and Renal Cell Cancer (HLRCC): Clinical, molecular and surveillance features in a cohort of 185 affected individuals. <b>Claire Forde.</b>	Prenatal exome sequencing in unexplained fetal oedema: a game changer. <b>Esther Dempsey.</b>
14:15	Structural Aberrations with Secondary Implications (SASIs): consensus recommendations for reporting of cancer susceptibility genes identified during analysis of Copy Number Variants (CNVs). <b>Sabrina Talukdar.</b>	Early onset polycystic kidney disease associated with functionally significant PKD1 hypomorphic variants. <b>Miranda Durkie.</b>
14:20	Molecular diagnosis of mosaic disease in neurofibromatosis type I. <b>Emma Burkitt Wright.</b>	De novo missense variants in WDR37 associated with syndromic coloboma; findings from the 100,000 genomes project. <b>Eleanor Hay.</b>
14:25	BRCA1/2 genetic testing in a series of women with Uterine Serous Cancer. <b>Rachel Jones.</b>	Null variants and deletions in BRWD3 cause an X-linked syndrome of mild-moderate intellectual disability, macrocephaly, and obesity: a series of 17 patients. <b>Philip Ostrowski.</b>
14:30	Direct-to-consumer genetic test results: cautionary tales. <b>Rachel Horton.</b>	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. <b>Hannah Robinson.</b>
14:35	Parent and health professional experiences and views of genome sequencing for rapid diagnosis in critically ill children. <b>Melissa Hill.</b>	Clinical and genetic variability in children with partial albinism. <b>Patrick Campbell.</b>
14:45 – 15:10	<b>Afternoon Tea and Trade Exhibition</b>	

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

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

<b>15:10 – 17:30</b>	<b>Applications of Genomics in Management of Human Disease (Wolfson Theatre)</b>	
15:10	Experience as knowledge: reproductive decisions of families living with genetic disease.	<b>Felicity Boardman</b> ( <i>University of Warwick</i> )
15:40	Lessons and opportunities from curating the genotypes of half-a-million people in the UK Biobank.	<b>Clare Bycroft</b> ( <i>formerly UK Biobank</i> )
16:10	Finding the origins of childhood cancer	<b>Sam Behjati</b> ( <i>Sanger Centre</i> )
<b>16:40</b>	<b>The BSGM Lecture 2019</b>	
	From syndromology to population cancer risk stratification: a life's journey in cancer genetics	<b>Professor Gareth Evans from The University of Manchester</b>
<b>17:30 – 18:00</b>	<b>BSGM Annual General Meeting</b>	