

British Society for Genetic Medicine Annual Conference 2026

20 November 2026 | 08:30-18:00 | Royal College of Physicians, London

Agenda

- 08:30** **Registration and refreshments**
Sponsor Exhibition
- 09:00** Welcome
Dr Lynn Greenhalgh, BSGM Chair
- 09:05** Opening Lecture
Professor Anneke Lucassen, University of Oxford
- 09:30** **Session 1:** Population resources and applications - chaired by Dr Harry Leitch & Associate Professor Vanessa Sancho Shimizu
*09:30-09:50 Professor Damian Smedley, '**Computational genomics approaches to rare disease diagnostics and disease gene discovery**', Queen Mary University of London*
*09:50-10:10 Professor Clare Turnbull, '**Navigating pathogenicity, penetrance and clinical utility for cancer susceptibility gene testing**', Institute of Cancer Research*
*10:10-10:30 Dr Julia Carrasco-Zanini-Sanchez, **title TBC**, Queen Mary University of London*
*10:30-10:50 Professor James Ware, **title TBC**, Imperial College London*
- 10:50** **Refreshment break, poster viewing & sponsor exhibition**
- 11:20** **Session 2:** VUS - chaired by Dr Charlotte Alston & Dr Hannah Titheradge
*11:20-11:40 Associate Professor Vanessa Sancho Shimizu, **title TBC**, Imperial College London*
*11:40-12:00 Dr Elizabeth Radford, **title TBC**, University of Cambridge*
*12:00-12:20 Professor Diana Baralle, **title TBC**, University of Southampton*
*12:20-12:40 Dr Melody Tabiner, **title TBC**, GenQA*
- 12:40** **Networking Lunch**

- 13:30** **Session 3:** ECR panel discussion, navigating career paths – chaired by Elizabeth Scotchman & Bushra Mussa
Frans Campher, Integral Leadership Dynamics
Dr Miranda Durkie, Sheffield Children's NHS Foundation Trust
Dr Laura Boyes, Birmingham Women's & Children's NHS Foundation Trust
Dr William Macken, Great Ormond Street Hospital for Children NHS Foundation Trust
- 14:10** **Session 4:** Selected abstracts I – chaired by Elizabeth Scotchman & Demetra Georgiou
14:10-14:22 – Matthew Heywood, '**Analysis of compound heterozygosity in aminoacyl-tRNA synthetase genes identifies missed diagnoses due to hypomorphic variants**'
14:22-14:34 – Sarah Hunt, '**Gene2Phenotype: accelerating diagnostic variant prioritisation with high quality, detailed gene-disease associations**'
14:34-14:46 – Emily Rees, '**Exploring what matters most to parents of children enrolled in clinical trials for rare metabolic conditions**'
14:46-14:58 – Sophie Allen, '**A novel likelihood-ratio methodology for combining case-control analyses (PS4) under reduced penetrance hypotheses within the up**'
14:58-15:10 – Dr Isaac Rothman, '**Uncovering Novel Gene Variants in Dystonia**'
- 15:10** **Refreshment break, poster viewing & sponsor exhibition**
- 15:40** **Session 5:** Selected abstracts II – chaired by Melissa Hill & Bushra Mussa
15:40-15:52 – Dr Charlie Rowlands, '**CanVar-UK: a digital platform supporting variant classification via an international diagnostic discussion forum**'
15:52-16:04 – Susan Walker, '**Increasing the diagnostic yield for families with rare disorders through Diagnostic Discovery**'
16:04-16:16 – Dr Harriet Cullen, '**The Huntingtin CAG repeat is a continuous modifier of brain structure and health vulnerability**'
16:16-16:28 – Martin Vu, '**Implementing genomic newborn screening in the NHS: what do healthcare professionals think?**'
16:28-16:40 – Karoline Kuchenbaecker, '**Equity in genome sequencing for rare disease diagnosis: lessons from the 100,000 Genomes Project**'
- 16:40** BSGM Lecture – chaired by Associate Professor Vanessa Sancho Shimizu
Professor Yanick Crow, University of Edinburgh, IMAGINE Institute
- 17:10** Awards & meeting summary
- 18:00** Networking reception
- 20:00** **Close**