BSGM
The British Society for Genetic Medicine

Annual conference

5TH NOVEMBER 2024

Royal College of Physicians, London

Conference Programme

#BSGM2024

09:00 - 09:55 ***Registration*** *(Wolfson Theatre foyer)*
Refreshments and sponsor exhibition *(Osler and Long Rooms, First Floor)*

Plenary session: Opening Lecture (Wolfson Lecture Theatre)

09:55 **Welcome** Demetra Georgiou *(Chair, BSGM)*

10:00 **Mendelian Genetics: An Archimedean lever in the era of AI**
Andrew Jackson *(University of Edinburgh)*

Plenary session: Big data and AI (Wolfson Lecture Theatre)

Chairs: Konrad Dobschuetz, Joo Wook Ahn

10:30 **AI based personalised drug and cell therapies in precision oncology and ultra rare conditions** Stephen Gilbert *(Technical University of Dresden)*

10:50 **AI ethics** Yves Moreau *(University of Leuven)*

11:10 **Utility of RNAseq to interpret and detect cryptic SVs**
Alistair Pagnamenta *(University of Exeter)*

Questions and discussion

Plenary session: Gene therapies BSGM/BSGCT (Wolfson Lecture Theatre)

Chairs: Ellie Chilcott, Flora Joseph

11:40 **Clinical application of gene therapy for rare and common diseases**
Rafael J Yáñez-Muñoz *(President, British Society for Gene and Cell Therapy)*

12:00 **Prenatal therapy for sickle cell disease** Panicos Shangaris *(King's College London)*

Questions and discussion

12:30 - 13:30 ***Lunch and sponsor exhibition*** *(Osler and Long Rooms, First Floor)*

Corporate Satellite Talk—ALEXION (Dorchester Library)

12:55 - 13:30 **Developments in Neurofibromatosis Type 1—An update for Geneticists**
Adam Shaw, Emma Burkitt-Wright

Concurrent submitted abstract sessions

Genetic spectrum of rare diseases

Dorchester Library, First floor

Chairs: Miranda Durkie, Verity Hartill

13:30 **AI-driven rare disease phenotype representation with application to diagnostics**
Michael Yates (*University of Edinburgh)*

13:40 **An Audit for Whole Genome Sequencing of 8000 diagnostic cases as part of the Genomic Medicine Service**
Gavin Ryan (*Regional Genetics Laboratory, Birmingham)*

13:45 **Functional genomics supporting diagnosis and therapy in phenylketonuria (PKU) – A national cohort study**
Rachael E McNeilly *(Bristol Genetics Laboratory)*

13:50 **Biallelic UGGT1 gene variants cause a congenital disorder of glycosylation**
Laura Harrold *(University of Exeter Medical School)*

14:00 **Leveraging natural language processing for a service improvement model in familial thoracic aortic aneurysm**
Alexander Deng *(Guy's and St Thomas's NHS Foundation Trust)*

14:05 **Keeping up with new gene discoveries: A simple computational approach for re-analysis of genomic data**
Fiona Price-Kuehne *(University College London Institute of Child Health)*

14:10 **Elucidating the clinical and genetic spectrum of inositol polyphosphate phosphatase INPP4A-related neurodevelopmental disorder**
Lettie Rawlins *(University of Exeter)*

14:15 Questions

14:30 **Rare disease genomic testing in the UK and Ireland: promoting timely and equitable access**
Emma Baple *(ACGS Rare Disease Position Statement Working Group)*

14:40 **Evaluation of a novel genetic testing pathway for inherited cardiac conditions through the GENetic testing VIDeo (GENVID) study: a revolution or evolution?**
Lisa Bryson *(West of Scotland Clinical Genetics Service, Glasgow)*

14:45 **UK population primary care electronic health record databases are valuable but underutilised resources for studying rare genetic conditions**
Thomas Wright *(Manchester University NHS Foundation Trust)*

14:50 **Are we effectively supporting parents making decisions about whole genome sequencing in the Genomic Medicine Service? A multisite survey study**
Ria Patel *(University College London Medical School)*

Cancer and new technologies

Wolfson Lecture Theatre

Chairs: Helen Hanson, Terri McVeigh

13:30 **The NHS Jewish BRCA-Testing programme: review of program data after first year reveals higher pathogenic variant detection rate within males.**
Bethany Torr *(Institute of Cancer Research, London)*

13:35 **Redefined indel classification framework reveals insights into mutational signatures of post-replicative repair deficiency**
Gene Koh *(Early Cancer Institute, Cambridge)*

13:45 **Mutational footprints of DNA damage and repair in homologous deficient human cells**
Salome Jingchen Zhao *(University of Cambridge)*

13:50 **In vitro assessment of chemotherapeutic drug Pidnarulex (CX-5461) reveals its extensive mutagenicity in human cellular models.**
Soraya Boushaki *(University of Cambridge)*

13:55 **Economic evaluation of extended panel analysis in cancer patients with historical NHS diagnostic germline genetic testing – A modeling study based on real-world data**
Qin Xi *(University of Cambridge)*

14:05 **Investigating sarcoma susceptibility through large-scale meta-analysis of case-control germline genomic sequencing data from multiple international data-sources**
Alice Garrett *(Institute of Cancer Research, London)*

14:10 **The shifting landscape of Fanconi anaemia: Insights from the UK registry with implications for clinical management**
Ramsay Bowden *(University of Cambridge)*

14:15 Questions

14:30 **The barriers to participation in genetic research experienced by three under-represented communities**
Rose Hall *(Royal Marsden NHS Trust)*

14:40 **Royal College of Physicians census: summarising two years of specialty-specific Clinical Genetics data**
Melody Grace Redman *(Yorkshire Regional Genetics Service)*

14:45 **What does a consent conversation for whole genome sequencing look like? an observational study of consent appointments in the NHS Genomic Medicine Service in England**
Holly Ellard *(UCL Great Ormond Street Institute of Child Health)*

14:50 **Use of Health Data and AI for Rare Disease Case Finding: Public Perception Survey Results**
Peter Fish *(Mendelian Ltd, London)*

(Continued over)

Concurrent submitted abstract sessions (continued)

14:55 **RTN4IP1 is essential for the final stages of mitochondrial complex I assembly and coenzyme Q biosynthesis**
Robert W. Taylor *(Newcastle University)*

15:05 **Adaptive long-read sequencing reveals a novel GGC repeat expansion disorder underlying spinocerebellar ataxia type 4**
Zhongbo Chen *(The Francis Crick Institute, London)*

15:15 Questions

14:55 **The role of genetic and genomic assistants, associates and practitioners (GenPs) in the NHS in England: Demographics, future career plans and challenges.**
Bethany Lumborg *(GenPAN Network)*

15:00 **DRAGENv4.0 increases the accuracy and utility of the Genomics England Rare Disease Bioinformatics Pipeline applied in the NHS Genomic Medicine Service**
Jamie Ellingford *(Genomics England Ltd, London)*

15:05 **Predicting: the future of health? Assessing the potential, risks and appropriate role of AI-powered genomic health prediction in the UK health system**
Harry Farmer *(Ada Lovelace Institute, London)*

15:15 Questions

15:30 - 16:00 ***Afternoon Tea and Sponsor Exhibition*** *(Osler and Long Rooms, First Floor)*

Plenary session: Developments in Genomics—National effort *(Wolfson Theatre)*

Chair: Sarah Bowdin

16:00 **Panel discussion**
Sue Hill *(Chief Scientific Officer & Senior Responsible Officer for Genomics—England)*
Catherine Ross *(Chief Scientific Officer—Scottish Government)*
Leon Wong *(Deputy Director, Health Science—Welsh Government)*
Ian Young *(Chief Scientific Advisor—Northern Ireland)*

17:10 ## The BSGM Lecture 2024

Chair: Demetra Georgiou

The best blood test I ever had
Sian Ellard *(University of Exeter Medical School)*

Awards and closing remarks

17:45 **Chair: Demetra Georgiou**

Networking event *(Osler and Long Rooms, First floor)*

18:00-20:00 Food and drink provided