

Course Programme

| Pre-course | | Introduction to genomics - genetics variation |
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| <i>Aim of session: Understand the fundamental concepts of genetic variation, gene expression, and regulation.</i> | | |
| Video | 20 mins | Introduction to the human genome <i>Andrew Read, University of Manchester, UK</i> |
| Video | 15 mins | Gene expression and regulation - Analysis of the transcriptome <i>Hans Hennies, University of Huddersfield, UK</i> |
| Video | 13 mins | Variation in the human genome - extent and source of variation <i>Sara Brown, University of Edinburgh, UK</i> |
| Video | 17 mins | Variation in the human genome, part II - How can we make sense of this? <i>Sara Brown, University of Edinburgh, UK</i> |
| Start (GMT) | Finish (GMT) | Presenter details |
| Monday 11 November 2024 | | |
| 12:00 | 13:00 | Registration, lunch and networking |
| 13:00 | 13:10 | Welcome Wellcome Connecting Science: <i>Nagehan Ramazanoglu Bahadir - Wellcome Connecting Science, UK</i> Scientific Programme Committee: Ajoy Bardhan, University of Birmingham, UK Joanna Jackow-Malinowska, Kings College London, UK David Kelsell, Queen Mary University of London, UK Edel O'Toole, Queen Mary University of London, UK |
| 13:10 | 14:00 | Introductions (ice-breaker activity) |
| 14:00 | 14:30 | Quiz on pre-course material <i>Chair: Joanna Jackow-Malinowska, Kings College London, UK</i> <i>Ajoy Bardhan, University of Birmingham, UK</i> |
| Comfort break | | |
| 14:40 | 16:20 | Session 1: Repair mechanisms and cancer <i>Aim of session: Explore the mechanisms of DNA repair in skin cells and their implications for cancer</i> <i>Chair: David Kelsell, Queen Mary University of London, UK</i> |
| 14:40 | 15:25 | Dermatlas: The Genomic Atlas of Dermatopathology David Adams, Wellcome Sanger Institute, UK |
| 15:25 | 16:10 | Genomics and DNA repair disordersfrom the lab to clinical applications Sophie Momen, St John's Institute of Dermatology, UK |
| 16:10 | 16:20 | Session discussion |
| 16:20 | 16:50 | Refreshment break and networking |

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| 16:50 | 18:30 | Session 2: Epigenetics |
| | | <i>Aim of session: Identify epigenetic mechanisms and non-coding genomic elements and their functions in skin biology.</i> <i>Chair: Edel O'Toole, Queen Mary University of London, UK</i> |
| 16:50 | 17:10 | Role of histone modifications in genome regulation Pradeep Madapura, Queen Mary University of London, UK |
| 17:10 | 17:30 | RNA-based mechanisms orchestrating the cell cycle and genome stability in cancer Lovorka Stojic, Barts Cancer Institute, UK |
| 17:30 | 18:30 | Workshop |
| 18:30 | 19:30 | Poster session |
| | | <i>Aim of session: Showcase and discuss your recent research findings and advancements in skin biology</i> Bar open (card payments only) |
| 19:30 | | Dinner |
| 19:30 | | Bar open (card payments only) |

Tuesday 12 November 2024

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| 09:30 | 11:00 | Session 3: Genomic literacy (workshop) |
| <i>Aim of session: Learn to perform basic variant interpretation using clinical case histories and genetic reports</i> | | |
| <i>Chair: David Kelsell, Queen Mary University of London, UK</i> | | |
| 09:30 | 11:00 | Genetic variation in clinical care - tools, tips and pitfalls? Elizabeth Jones, Saint Mary's Hospital Manchester and University of Manchester, UK Thomas Cullup, Great Ormond St Hospital, UK |
| 11:00 | 11:45 | Refreshment break and networking |
| 11:45 | 13:25 | Session 4: Genomic technologies in skin disease |
| <i>Aim of session: Explore advanced techniques and applications of long read sequencing and transcriptomic studies in skin research</i> | | |
| <i>Chair: Joanna Jackow-Malinowska, Kings College London, UK</i> | | |
| 11:45 | 12:30 | Long read sequencing for skin research John Common, A *STAR Skin Research Labs, Singapore |
| 12:30 | 13:15 | Transcriptomic studies of mouse paw skin in health and disease Diana Blaydon, Queen Mary University of London, UK |
| 13:15 | 13:25 | Session discussion |
| 13:25 | 14:25 | Lunch and networking |
| 14:25 | 15:45 | Session 5: Rare disease patient voice |
| <i>Aim of session: Understand and integrate patients' expectations for new treatments</i> | | |
| <i>Chair: Edel O'Toole, Queen Mary University of London, UK</i> | | |
| 14:25 | 14:50 | Scales of the Unexpected Mandy Aldwin-Easton, Ichthyosis Support Group (ISG), UK |
| 14:50 | 15:15 | Pain, isolation, and the life-changing work of PC Project Janice Schwartz, Pachyonychia Congenita Project, USA- VIRTUAL |
| 15:15 | 15:45 | Panel discussion |
| 15:45 | 16:15 | Refreshment break and networking |
| 16:15 | 17:05 | Session 6: AI and genomics |
| <i>Aim of session: Explore the potential of AI in skin research</i> | | |
| <i>Chair: Ajoy Bardhan, University of Birmingham, UK</i> | | |
| 16:15 | 17:00 | Generative machine learning to model cellular perturbations Mo Lotfollahi, Wellcome Sanger Institute, UK |
| 17:00 | 17:05 | Comfort Break |
| 17:05 | 18:05 | Session 7: Keynote speaker |
| <i>Chair: Joanna Jackow-Malinowska, Kings College London, UK</i> | | |
| 17:05 | 18:05 | Human skin in health and disease: lessons from genomics Muzlifah Haniffa, Wellcome Sanger Institute, UK |
| 18:05 | 19:05 | Poster session with drinks reception |
| <i>Aim of session: Showcase and discuss your recent research findings and advancements in skin biology</i> | | |
| 19:05 | | Dinner |
| 19:05 | | Bar open (card payments only) |

Wednesday 13 November 2024

09:30 10:30 Session 8: Therapies (keynote speaker)

Aim of session: Understand the significance of genetic diagnosis in skin disease and its role in developing effective therapies

Chair: Joanna Jackow-Malinowska, Kings College London, UK

09:30 10:30 Gene therapy for epidermolysis bullosa
[Peter Marinkovich, Stanford University, USA](#)

10:30 11:00 Refreshment break and networking

11:00 12:00 Session 9: Therapies (continued)

Aim of session: Understand the significance of genetic diagnosis in skin disease and its role in developing effective therapies

Chair: Edel O'Toole, Queen Mary University of London, UK

11:00 11:40 Unravelling sporadic vascular malformations due to mosaic RAS/MAPK variants: new therapeutic perspectives
[Maanasa Polubothu, University College London, UK](#)

11:40 12:20 Therapeutic Gene Editing Technologies in Dermatology
[Joanna Jackow-Malinowska, Kings College London, UK](#)

12:20 12:30 Session discussion

12:30 12:50 Consolidation and Closing remarks

Scientific Programme Committee:

[Ajoy Bardhan, University of Birmingham, UK](#)

[Joanna Jackow-Malinowska, Kings College London, UK](#)

[David Kelsell, Queen Mary University of London, UK](#)

[Edel O'Toole, Queen Mary University of London, UK](#)

12:50 13:00 Grab bag lunch

13:10 Coach departures for Cambridge train station and city centre